985

1985 Catalog of Cell Lines

NIGMS Human Genetic Mutant Cell Repository

NIA Aging Cell Repository

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Cover Photograph: Spread of human chromosomes treated with bromodeoxyuridine to produce a replication R banding pattern and to highlight the late replicating X chromosome (unstained). Courtesy of Dr. Margaret M. Aronson, Institute for Medical Research.

1985 Catalog of Cell Lines

NIGMS Human Genetic Mutant Cell Repository
sponsored by the
National Institute of General Medical Sciences

NIA Aging Cell Repository
sponsored by the
National Institute on Aging

Location of Repositories:
Institute for Medical Research
Copewood and Davis Streets
Camden, N.J. 08103
(609) 966-7377

U.S. DEPARTMENT OF HEALTH
AND HUMAN SERVICES
Public Health Service
National Institutes of Health

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Announcement of Fees

The fees for a 25 cm^2 flask of cells shipped before December 31, 1985 are:
$50 per flask for academic, non-profit institutions

$100 per flask for commercial, industrial, for-profit establishments

The fees for cells shipped after this date are $60 and $120 respectively. Orders for 75 or more cell cultures placed at the same time will receive a 50% discount of $30 per culture. The quantity discount does not apply to purchases made by individuals from for-profit institutions.

Shipping charges are not included in these fees but are added to the invoice at the time of billing. These charges vary depending upon the mode of shipment requested by the purchaser.

Catalog Requests

Additional copies of this catalog are available, free of charge, by contacting the Repository.

PREFACE TO THE TWELFTH EDITION

Since its inception in 1972, the Human Genetic Mutant Cell Repository has processed more than 6,500 submitted cell cultures, tissue biopsies, and peripheral blood specimens and provided more than 30,000 cell cultures to investigators. These activities have resulted in a worldwide reputation for excellent, well characterized, thoroughly documented, and contaminant-free cell cultures.

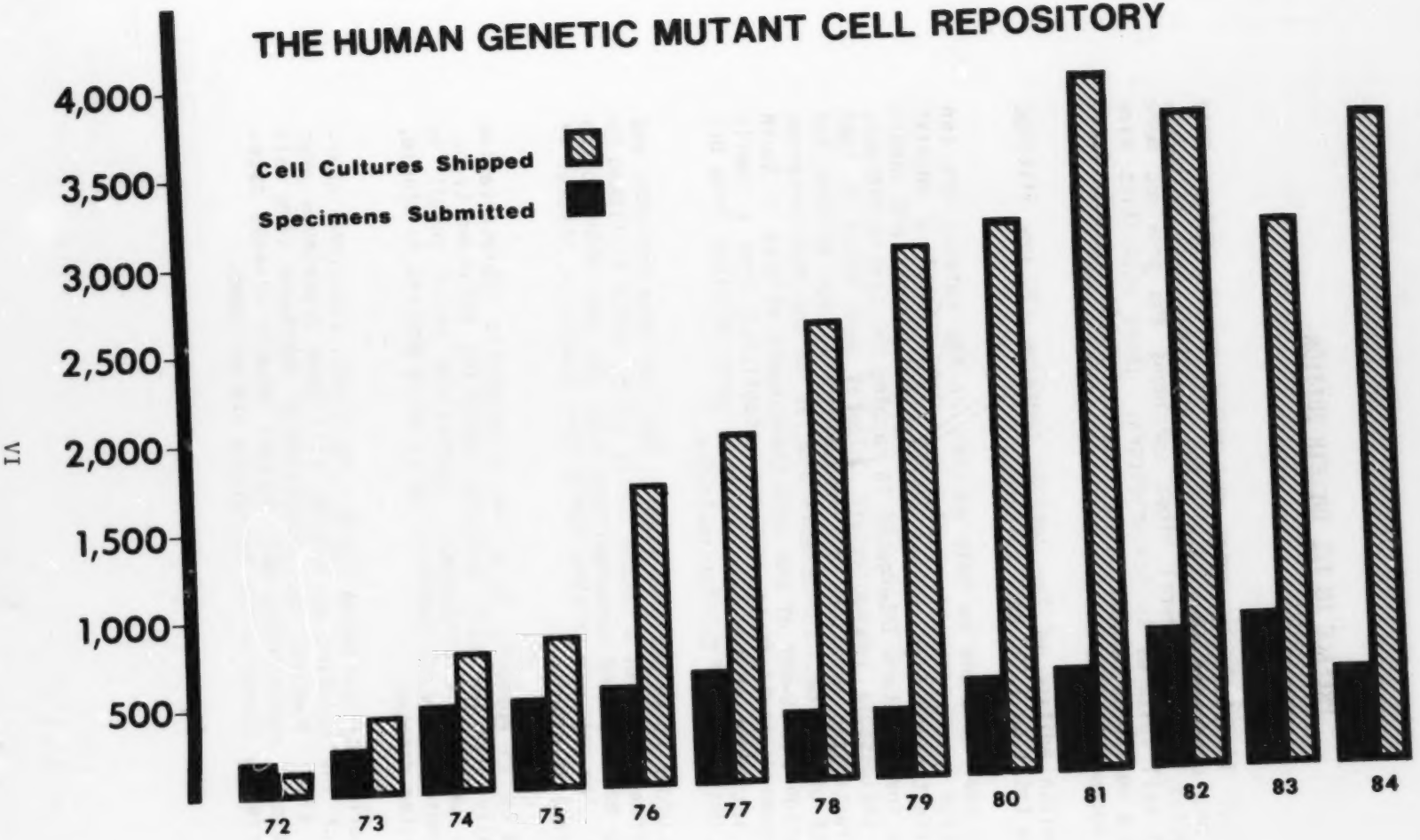
The twelfth edition of the catalog contains 290 new listings bringing the total to 3,590 cell cultures.

Special new additions to this edition of the catalog are ten multigeneration cystic fibrosis pedigrees. These families consist mainly of affected siblings, normal siblings and parents and should be useful to investigators interested in mapping the cystic fibrosis gene. Nine thalassemia lymphoblastoid cultures were added in 1984 and the characterized mutations of the collection now account for more than 95% of the beta-thalassemia alleles in the Mediterranean basin population and 80-85% of the beta-thalassemia alleles in South China. Thirteen lymphoblastoid cultures established from a family with a high incidence of tuberous sclerosis were obtained from Dr. Moyra Smith, University of California-Irvine.

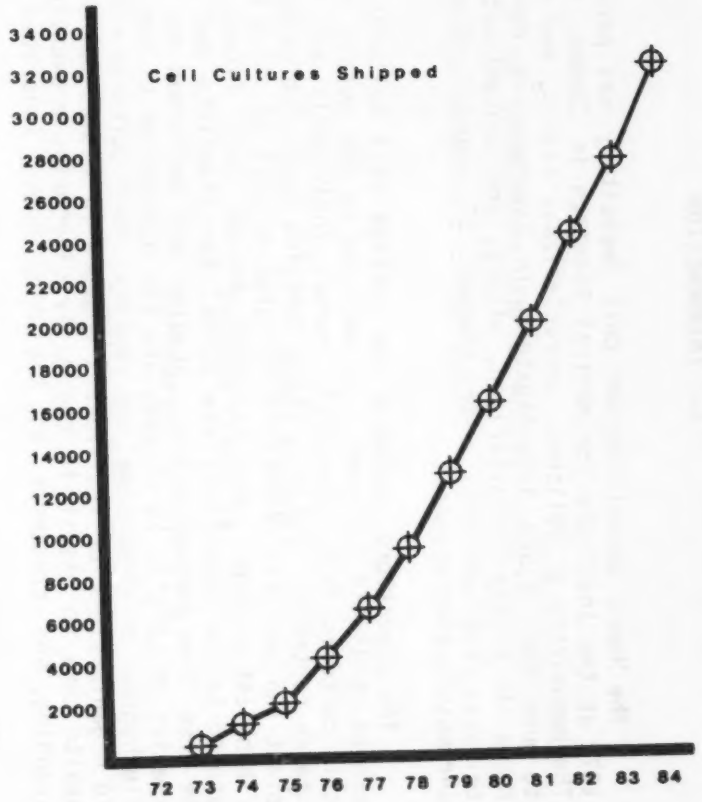
The Old Order Amish pedigree (family 884) has been expanded and 12 cultures from members of a second family (1075) which is linked to family 884 by marriage and consanguineous ties has been added to the 1985 catalog. The patients in this family have bipolar I, bipolar II or major depressive disorders.

Included as an Appendix is a new diagrammatic representation of chromosomally aberrant cell cultures depicting balanced translocations and unbalanced chromosomal aberrations which result in specific chromosome segments (whole or part) being present in single, triple or greater dosages.

As the collection approaches 7,000 submissions, additional characterization of the existing collection will take precedence over expanding its size. However, the submission of specimens from well documented patients representing well defined genetic disease categories not currently stored in the Repository are welcomed.


THE HUMAN GENETIC MUTANT CELL REPOSITORY
Cell Cultures Shipped
Specimens Submitted
4,000
3,500
3,000
2,500
2,000
1,500
1,000
500
72
73
74
75
76
77
78
79
80
81
82
83
84

NIGMS Human Genetic Mutant Cell Repository
NIA Cell Culture Repository
CUMULATIVE TOTALS



I. INTRODUCTION

The Human Genetic Mutant Cell Repository was established in 1972 at the Institute for Medical Research in Camden, New Jersey. The Repository establishes, characterizes, stores, and distributes to more than 2,500 investigators in laboratories doing genetic research, highly characterized, viable, and contaminant free cell cultures from patients with biochemical genetic disorders and chromosome aberrations.

The Repository contains low passage cell cultures which are stored in liquid nitrogen. In addition to the mutant cell lines, cell cultures from apparently normal individuals are stored for use as controls. The collection contains human fibroblast, lymphoblast and amniotic fluid cell cultures as well as virus transformed fibroblast cultures and selected animal cell lines. Cultures stored in the Repository are tested for viability and assured to be free from contamination. Studies are performed to verify the species of origin, karyotype, and the expression of the particular biochemical or chromosomal abnormality. Cell cultures are available to organizations or individuals engaged in biomedical research or health care to assist in the early diagnosis, prevention, understanding, and treatment of the many human inherited diseases.

The staff of the Repository are proud of their contributions to the application of cell culture techniques to the detailed study of human genetics which have led to a wealth of information about metabolic disorders and gene abnormalities. This highly specialized program has been developed and maintained under the supervision of the NIGMS project officer, many consultants, and a scientific advisory committee which periodically reviews the progress of the Repository and makes specific recommendations related to the acquisition of new cell lines, deletion of cell lines from the collection and catalog format changes. Appreciation is also expressed to the many investigators who submit interesting specimens to the Repository and to those researchers who serve as curators by analyzing cell cultures for specific biochemical and chromosomal abnormalities.

Interested investigators are encouraged to utilize the Repository as a source of verified human genetic mutant cell cultures.

Arthur E. Greene, D.Sc., Director

Richard A. Mulivor, Ph.D., Associate Director

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Elizabeth Hoffman, B.S.

Roger Yu, M.S.

II. ORDERING CELL CULTURES

1. REQUIREMENTS

The cells are distributed only to qualified professional persons who are associated with recognized research, medical, educational or industrial organizations engaged in health-related research or health delivery. Before cell cultures can be shipped, the assurance form (Page XII) must be signed and returned to the Repository. This is adequate for most cell cultures in the Repository. However, lymphoblast or virus-transformed cell cultures require prior agreement to observe the Minimum Safety Guidelines (Page XVII) and return of the signed Agreement on Lymphoid and Virus-Transformed Cells (Page XIV). In addition, certain cell lines require a compliance form (Page XVI) before they can be shipped. These forms may be obtained by writing to the Repository, or may be photocopied from the catalog.

2. PROCEDURE

Requests for cell cultures must be submitted on institutional purchase forms including purchase order numbers. Purchasing agents should indicate the name of the investigator on such orders. Telephone orders will be accepted when accompanied by a purchase order number and should be confirmed within several days by letter. Each culture requested should include the Genetic Mutant Repository number (Rep.#) and the diagnosis. When an investigator wishes to order both established cultures and frozen biopsies which must be established as cultures before they can be supplied, separate purchase orders for each type of sample must be submitted.

3. ADDRESS

All requisitions should be addressed to:

Dr. Arthur E. Greene
The Human Genetic Mutant Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103

Telephone 609-966-7377

4. FEES

The fee for a 25 cm^2 flask of cells shipped before December 31, 1985 is $50 per flask for academic, non-profit institutions and $100 per flask for commercial, industrial, for-profit establishments. After this date, the fees will be $60 and $120 respectively. Orders for 75 or more cell cultures placed at the same time will receive a 50% discount of the full price or $30 per culture. This reduction does not apply to purchases made by individuals from for-profit establishments. Frozen ampules are not shipped, because experience

has shown much better success in shipping freshly recovered growing cultures. Mass production of cell cultures is not a function of the Repository. The Repository is designed to provide only seed cultures. It is suggested that the recipient store aliquots of cells in early passage in liquid nitrogen as insurance against contamination, accidents, artifacts associated with aging, and loss of the culture.

5. SHIPPING CHARGES

The shipping and packaging charges are prepaid and are added to the invoice at the time of billing. Shipping charges vary depending upon the mode of shipment requested by the purchaser.

6. HOW SHIPPED

All cell cultures are grown and frozen in antibiotic-free media to aid in the detection and prevention of contamination. Cell cultures in the Repository have been tested and found free of mycoplasma, bacteria, and fungi during characterization, at the time of frozen storage, and after recovery from liquid nitrogen. When an order is received, a frozen ampule is usually recovered from liquid nitrogen on the following Thursday. The culture is inspected for growth and contamination and the medium is changed on Friday. The following Monday, the culture is inspected, the medium is removed and the T25 flask is filled with fresh medium, packed and shipped by air mail special delivery. The flask is enclosed within two watertight plastic envelopes within a styrofoam box in a cardboard mailer to prevent leakage, overheating or freezing during shipment. The only exception to this schedule occurs when a frozen biopsy is ordered. All requests for frozen biopsies will be reviewed for acceptance. If the order is to be filled, the biopsy will be recovered from liquid nitrogen, established in culture, and tested for contamination before it will be shipped. There will be a 2 month time period needed to process these orders. Upon receipt, cell cultures should be placed in the incubator at 37°C for a few hours or overnight to permit recovery from damage which may have occurred during shipment. Return postcards are enclosed. Please return the postcards with notations about the condition on arrival and planned utilization. Pertinent suggestions to improve the effectiveness of the Repository are appreciated. Over the past 5 years, 94% of shipments have arrived in satisfactory condition.

7. DESCRIPTIVE DATA

Each shipment includes a computer generated information sheet describing the cell culture, suggested directions for cell growth, and pertinent references when available.

8. REQUEST FOR CITATION AND REPRINTS

It will be greatly appreciated and will make the cell collection more valuable if the source and Repository number are cited in publications in which cell cultures from the Human Genetic Mutant Cell Reposi-

tory are used. The Repository would appreciate receiving a reprint of such publications.

9. PUBLICATION OF DESCRIPTIVE ARTICLES ON CELL CULTURES STORED IN THE REPOSITORY

Concise descriptions of some of the cultures stored in the Repository are published after characterization in Cytogenetics and Cell Genetics under the name of the individual originally submitting the culture or biopsy. These descriptions include brief clinical histories, assay methods when applicable, family pedigrees, full or partial karyotypes in the case of chromosomal aberrations, and references to previous work involving the cell lines. For a listing of these and other papers see Appendix C, page 490.

10. ASSURANCE

Before cells can be shipped from the Human Genetic Mutant Cell Repository, the recipient institution must submit a purchase order number and agree to the following limitations. This agreement must be renewed annually. Please sign and return original to the Institute for Medical Research, Copewood Street, Camden, New Jersey 08103.

ASSURANCE FORM

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM4-2100, National Institute of General Medical Sciences.

It is further agreed that cell cultures obtained from the Human Genetic Mutant Cell Repository will not be resold, but they may be replicated by a third party for the original purchaser. The third party shall not be allowed to resell other than to the original purchaser furnishing the material for replication and the third party shall not use such cells for human experimentation.

Name of Institution	Name of Principal Investigator	Date
Signature of Department Chairman	Signature of Principal Investigator	Date

11. AGREEMENT ON LYMPHOID AND VIRUS-TRANSFORMED CELLS

This document must be appropriately completed and exchanged before lymphoid and virus-transformed cell lines can be transferred from the Human Genetic Mutant Cell Repository to your laboratory.

AGREEMENT

The potential hazardous nature of these human cell lines is unknown. However, in view of this lack of knowledge, it is appropriate that the scientific community be made aware that a potential hazard may exist. Therefore, we recommend that the enclosed guidelines for laboratory procedure be adhered to in the handling of lymphoid and virus-transformed cell lines. They are also recommended for handling all cell cultures.

It is understood that you will not further distribute cell lines sent to you to laboratories not under your direct supervision. It is further agreed that all cultures obtained from the Human Genetic Mutant Cell Repository may only be replicated by a third party if the third party also executes an Agreement on Lymphoid and Virus-Transformed Cells agreeing to follow the Minimum Safety Guidelines.

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository you agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM4-2100, National Institute of General Medical Sciences.

This agreement, when appropriately executed, will be on file and will qualify you to receive lymphoid and virus-transformed cell lines from the Human Genetic Mutant Cell Repository.

AGREEMENT ON LYMPHOID AND VIRUS-TRANSFORMED CELLS

We the signatories below, have read and understand this document and agree that in the handling of cell lines furnished, we will adhere to the procedures and recommendations outlined in the foregoing and the attached "Minimum Safety Guidelines Recommended for Working with Lymphoid and Virus-Transformed Human Cell Lines".

A. For the recipient laboratory:

Investigator: Signature _____ Date _____

Title _____

Head of Laboratory or Immediate Supervisor: Signature _____ Date _____

Title _____

Address _____

12. COMPLIANCE AGREEMENT

Ownership of Special Cell Lines in the Cell Culture Repository

Contributors of cell lines have frequently invested much time, effort and expertise in their development. Some of these cell lines are patentable and therefore potentially of great economic value to the contributors. The new patent law, P.L. 96-517, passed in December, 1980, permits universities, small companies and non-profit organizations to retain ownership of patents gained as a result of federal grants and contracts. These new developments can greatly accelerate or greatly retard medical progress. It can accelerate the transfer of new findings to the marketplace if a commercial company can get exclusive rights to exploit the the results of non-profit research. This would justify commitment of development funds by the company. On the other hand, it can greatly retard research progress if investigators and nonprofit organizations are reluctant to fully exchange research findings and cell cultures because of pending patent applications or potential commercial rewards, or because there is no central place where verified cell cultures can be made available for research purposes. The Institute for Medical Research believes that the proper solution to these conflicts is to establish a special policy for the distribution of selected cells for research only. Title, in this case, will remain with the contributor. Each recipient of a cell culture must sign a waiver to use the cell cultures for research only and not to give or sell it to a third party for commercial purposes or for sale of cells or cell products. For any such commercial use whatever the

recipient of cells from the Repository must first seek and obtain written permission from the person who contributed the cell lines to the Repository.

Cell cultures requiring a compliance form have their Repository numbers preceded by a ➤ in the catalog.

COMPLIANCE FORM

Waiver and Prior Agreement Before Receipt of Repository Cell Culture #

"Ownership of cell line # in the repository resides with the original developer. No distribution of the cell line shall be made prior to signed agreement of this waiver. The cell line is provided for research purposes only, and it or its products shall not be sold or used for commercial purposes, nor will cells be distributed further to third parties for purposes of sale, or producing for sale cells or their products. Secondary distribution shall only be after written approval by the contributor of the cell line. The cells are provided as a service to the research community. They are provided without warranty of merchantability or fitness for a particular purpose or any other warranty, expressed or implied. In addition, the recipients of the cell lines agree to indemnify and hold harmless the United States government and IMR and the contributor from any claims, costs, damages, or expenses resulting from any injury (including death), damage, or loss that may arise from the use of the cell line."

Date_____

Signed _____
Name of Investigator

_____
Institution

Signed _____
Department Chairman

III. HUMAN GENETIC MUTANT CELL REPOSITORY

MINIMUM SAFETY GUIDELINES RECOMMENDED FOR WORKING WITH LYMPHOID AND VIRUS-TRANSFORMED HUMAN CELL LINES*

A. Supervision

1. Administrative Responsibilities

a. Responsibility of Management

Management should establish a biohazards committee to institute and enforce a health and safety policy which includes a specific safety program for work involving human cell lines. The program should meet applicable federal, state and local regulations and include safety training, maintenance of accident records, and provisions for emergency treatment.

b. Responsibility of the Principal Investigator

The principal investigator is responsible for the preparation of safety protocols for the research program under his direction. The protocols should include appropriate procedures for use, storage, decontamination, disposal and emergency treatment. The protocols should be approved by the biohazards committee and discussed with the research staff before starting the research program.

2. Medical Surveillance and Screening

a. Physical Examinations

Appropriate pre-employment and periodic medical examinations are desirable for persons working with human cell lines.

b. Work Restrictions

Persons having reduced immunologic competency should be restricted from working with these human cell lines.

*These guidelines were developed on February 11, 1974 at a special meeting at the National Institutes of Health attended by representatives from the Office of Biohazards of NCI, NIAID, NIGMS and the Advisory Committee to the Human Genetic Mutant Cell Repository.

c. Serum Collection

Serum should be collected at the time of the pre-employment physical to establish a baseline reference. Serum should be re-collected and stored annually. Serum should be collected immediately after accidental injection or ingestion and at an appropriate interval thereafter. For those individuals exposed to long term lymphoid lines or their derivatives, anti-EB virus titers should be obtained on the collected sera.

3. Laboratory Access

Access to the cell culture area should be restricted to persons directly working with the cell lines, or by specific authorization by the principal investigator or director of the laboratory.

B. Personnel Practices

1. Pipetting

Mechanical pipetting aids rather than mouth pipetting should be used for all pipetting procedures.

2. Eating, Drinking and Smoking

Eating, drinking and smoking should not be allowed in the same areas where cell lines are under study.

3. Protective Clothing

It is recognized that the criteria for protective clothing may vary according to the physical situation of the laboratory and the agents handled. Ideally, adequate protective clothing such as a fully fastened laboratory coat should be worn. This clothing should not be worn outside the work area once the work area has been entered.

C. Physical Control Practices (Recommended for all cell lines, but required for long term lymphoid lines and their derivatives).

1. Ventilated Safety Cabinets or Hoods

Ventilated safety cabinets and hoods and other safety apparatus should be employed and should be tested at least annually to certify correct containment and operation. A list of specifications for satisfactory hoods and instructional materials may be obtained from the Office of Biohazards, NCI.

2. Housekeeping

Appropriate housekeeping procedures which suppress the formation of aerosols should be used. Working surfaces should be wiped down

with an appropriate disinfectant before and after work with each cell culture and at the end of the working day.

3. Decontamination and Disposal

Contaminated glassware and similar materials should be appropriately decontaminated or stored for decontamination before removal from the work area for recycling or disposal. Liquid wastes should be decontaminated either chemically or by heat, before being discharged to the community sanitary sewer system.

4. Protection of Vacuum Lines

Vacuum services, if used, should be protected with disposable absolute air filters and liquid traps. The effluent should be collected in liquid traps containing concentrated disinfectant.

References

Biohazards in Biological Research, ed. A. Hellman, M.N. Oxman and P. Pollack. Cold Spring Harbor Laboratory, New York (1973).

IV. SUBMISSION OF SPECIMENS TO THE REPOSITORY

Biopsies and bloods are preferred to established cell cultures because they can be processed and stored in lower passage. When submitting a specimen to the Repository, the biopsy should be placed in a 25 cm^2 tissue culture flask or screw top vial with culture medium containing 100 units/ml of penicillin and 100 mcg/ml of streptomycin. Tape the top or cap of the flask very securely to prevent leakage. Bloods should be approximately 10-15ml in volume and contain heparin to prevent clotting. Package the specimen in a container so that it will not be broken in transit. Mark on the outside of the package that it should be kept at room temperature and not refrigerated, frozen or overheated.

Early passage cultures may be submitted when a biopsy or blood cannot be obtained from the patient. The culture flask (25 cm^2 preferably) should be filled to the top with culture medium and shipped as described above. Mail submission <u>prepaid</u> by air mail special delivery to:

Dr. Arthur E. Greene
The Human Genetic Mutant Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103

Phone 609-966-7377

Before a submission can be processed for inclusion in the Repository, clinical and laboratory documentation proof of the diagnosis must be provided on a <u>submission sheet</u> which is available from the Institute for Medical Research, or may be photocopied by the submitter from pages XXI-XXIV of this catalog. Without this information the specimen cannot be coded into the information retrieval system or be of value to users of the Repository. To conserve effort and expense, the Repository staff are instructed not to process a specimen unless documentation of diagnosis is provided with the specimen. Submitters are therefore requested to fill in <u>all</u> applicable blanks on the submission sheets and provide a family pedigree if available. If a chromosomally aberrant specimen is submitted, inclusion of a photograph of the karyotype is appreciated.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

The submitter of a specimen that is accepted for inclusion in the Repository may receive without charge a single sample of the cell line derived from this submission or a single sample of a different cell line.

TO BE FILLED IN BY IMR

Repository Number _____

Date Received ___/___/___
Mo. Day Year

SUBMISSION SHEET

Please Complete Every Item that Applies

Submitting Investigator _____

PATIENT INFORMATION:

SPECIES: HUMAN _____ OTHER _____

PATIENT'S INITIALS: _____ DATE OF BIRTH _____

AGE: _____ (circle one)
NB (newborn) DA (days) WK (weeks)
MO (months) YR (years)
If Fetal Tissue _____ FW (weeks of gestation)

PHENOTYPIC SEX: (circle one) M (male) F (female) A (ambiguous)

RACE: (circle one) BL (black) WH (white) AI (American Indian)
OR(oriental) IN (Asiatic Indian) Other _____

ETHNIC BACKGROUND (if relevant): _____

CLINICAL PHENOTYPE: A (clinically affected) C (clinically normal)
H (heterozygous) R (at risk) V (variant) O (other)
B (balanced chromosome variant) U (unbalanced chromosome variant)

DIAGNOSIS: _____

McKUSICK NUMBER _____

CLINICAL MANIFESTATIONS: (Please include detailed clinical reports when available) _____

_____

_____

SOURCE OF CLINICAL INFORMATION:

____ 1. Personal Examination
____ 2. Hospital Records
____ 3. Genetic Clinic Records
____ 4. Specialist's Report
____ 5. Autopsy Records
____ 6. Private Physician
____ 7. Other _____

IS A PEDIGREE INCLUDED: (circle one) Y (yes) N (no)
(Please include a detailed pedigree if it is available)

LABORATORY INFORMATION: Local I.D. Number _____

TYPE OF SAMPLE SUBMITTED: (circle one)

Culture
- C (fibroblast culture)
- L (lymphocyte culture)
- A (amniotic fluid cell culture)
- E (differentiated cell culture)
- Other _____

Tissue Sample
- B (skin biopsy)
- P (peripheral blood)
- Other _____

Passages of culture submitted _____

Biopsy Site _____

Population doubling level _____

Date culture was established _____

CULTURE MEDIA in which submitted:

McCOYS 5A	_____ %	HAMS F-10	_____ %
HANKS-EAGLES	_____ %	RPMI 1640	_____ %
EAGLES-EARLES	_____ %	OTHER	_____
HAMS F-12	_____ %	FETAL BOVINE SERUM	_____ %
		Inactivated	_____ I
		Uninactivated	_____ U

ADDITIVES ___Y___ (yes) ___N___ (no)

ANTIBACTERIAL
- ____ Penicillin
- ____ Streptomycin
- ____ Gentamycin
- ____ Other

ANTIFUNGAL
- ____ Fungizone
- ____ Mycostatin
- ____ Other

REQUIRED NUTRIENT
Specify _____

Special Instruction on growth, handling, or freezing: _____

ADDITIONAL PERTINENT INFORMATION:

Has this patient/family been reported in the literature: Y (yes) N (no)

Please cite reference(s): _____

For the disease category you are submitting, is the defect expressed in

Fibroblasts Y (yes) N (no)
Lymphoblasts Y (yes) N (no)

DIAGNOSTIC LABORATORY TESTS AND STUDIES

BIOCHEMICAL STUDIES: Y (yes) N (no)

Were the studies done on this culture? Y (yes) N (no)

If no, what tissue or culture? _____

TYPE OF ASSAY _____

_____

METHOD USED _____

_____ (please cite reference)

RESULTS FOR PATIENT? _____

RESULTS FOR CONTROLS? _____

RESULTS ARE: (circle one)

1. Consistent with Homozygosity
2. Consistent with Heterozygosity
3. Inconclusive
4. Normal levels
5. Other _____

CHROMOSOME STUDIES: Y (yes) N (no)

Were the studies done on this culture? Y (yes) N (no)

If no, what tissue or culture? _____

Current Paris Nomenclature _____

_____

The Chromosome variant is: B (balanced) U (unbalanced)

Staining methods used: _____

Other Chromosome analysis: _____

_____ increased breakage? _____ sister chromatid exchange?

Have other specimens from this patient/family been stored in the Repository in Camden? Y (yes) N (no)

If yes, what are their Repository numbers and what are their relationships: _____

_____

ADDITIONAL REMARKS:

_____

_____

_____

_____

RELEASE, PERMISSION AND CONSENT FORM

To encourage storage of unusual cell cultures in the Repository, provision has been made for delayed release to other investigators if the contributor so desires. Please check your preference: a) release culture to anyone requesting it _____, b) release only to contributor during the first year _____. At the conclusion of 1 year the cell culture will be listed in the next printing of the catalog and made available to other investigators unless the submitter obtains a delay from the project officer at NIGMS.

I hereby grant permission for these cells to be stored in a bank of genetic mutant cell cultures and the progeny cells distributed to qualified investigators. Appropriate consent was obtained from the patient from whom the cells were originally obtained, or can be reasonably inferred, for use of these cells for diagnosis, research, teaching or therapy.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

Date _____ Submitter _____
(Signature)

Address _____

_____

Telephone Number _____

Mail completed form with, or preferably preceeding, shipment of cell cultures, to: Dr. Arthur Greene, Institute for Medical Research, Copewood and Davis Streets, Camden, New Jersey 08103.

Telephone (609) 966-7377.

V. USE OF THE CATALOG

A. Locating a Cell Culture

The overall format of the catalog can be seen by consulting the Table of Contents (page I).

A cell culture belonging to a particular disease category may be found by consulting the alphabetical index of diagnoses comprising Appendix I, page 621. Following each diagnosis is the page number on which that disease may be found.

If the Repository number of a culture is known, its page location may be identified by consulting Appendix H, page 591 which lists Repository number versus page number. Fibroblast, Lymphoblast, and Amniotic Fluid cell cultures from the same individuals or family groups are cross referenced in the remarks column for each cell line.

B. Explanation of the Data Columns of the Catalog

COLUMN

REP #: The Repository number of the cell culture is the number assigned to it upon receipt at the Human Genetic Mutant Cell Repository. It consists of two letters and four digits, i.e., GM1234. Fibroblast cultures originate from skin unless otherwise noted. Lymphoblast cultures usually originate from peripheral blood. When it becomes necessary to expand the supply of an original cell line, the expansion is designated by adding a letter to the Repository number, i.e., GM1234A. The expansion differs from the original cell line in that its passage number may be higher. If a Repository number is followed by the letter Z, i.e., GM1234Z, it has been stored in the Repository as a frozen biopsy and has not been established as a culture. Workup of these specimens will be made as the demand warrants.

Passage #: The number of serial in vitro transfers of the cell culture as stored in liquid nitrogen. If the total number of passages is unknown, this column is left blank and the total number of passages at IMR is indicated in the remarks column. Passage numbers are not recorded for lymphoblast cultures.

Gen Time: The time required for a logarithmically growing lymphoblastoid culture to double its number of viable cells.

Culture Medium: For explanation of the code see Appendix A, page 469. This column describes the recommended growth medium and percentage of fetal bovine serum.

Submitter Code: A number which identifies the investigator who submitted the specimen to the Repository. For code and submitter names see Appendix B, page 470.

Age: The age of the donor at the time the specimen was obtained, is expressed in years or, when appropriate, in months (MO), weeks (WK), days (DA), or fetal weeks of gestation (FW).

Sex: Phenotypic sex; Male (M), Female (F), or Ambiguous (A)

Race: WH-Caucasian; BL-Black; OR-Oriental; IN-Indian (India); AI-American Indian; BR-Polynesian. Space is left empty if race unknown.

Genetic Status:

+	Normal gene
-	Mutant gene
Y -	Hemizygous for X-linked trait
+ ?	Genetic status not determined
+ -(0)	Carrier for trait as determined by pedigree or clinical diagnosis

The McKusick* number immediately following the name of the disease indicates the dominant, recessive, or X-linked nature of the disease. The first digit is one for a dominant disease (e.g. Huntington Chorea - 14310), two for a recessive disease (e.g. Tay-Sachs Disease - 27280), and three for an X-linked trait (e.g. Lesch Nyhan Syndrome - 30800).

Verified: The clinical diagnoses of all cell cultures listed in the catalog have been verified by the submitter and reviewed by the staff and one or more members of the Advisory Committee. B is entered in this column if the defect (biochemical or chromosomal) was verified on the cell culture before submission to the Repository; A is entered if verified after recovery of Repository cultures from liquid nitrogen. The space is left blank if the defect was verified on another cell culture or tissues from the same patient or relative, or if verification is not possible because the defect is unknown or not expressed in culture.

Paris Nomenclature: As described (Paris Conference, 1971 and Supplement, 1975): Standardization in Human Cytogenetics. Birth Defects: Original Article Series, VIII:7, 1972, and XI: 9, 1975. The National Foundation, New York.

* McKusick, Victor. Mendelian Inheritance in Man, 6th Edition. Johns Hopkins Press, Baltimore, 1983.

Balanced, Unbalanced: Balanced (B) or Unbalanced (U) karyotype

Relation to Proband: Example: AFF BRO = Affected brother.

Fam #: A unique pedigree number is assigned if cultures from more than one member of a family or more than one sample type (i.e. fibroblast and lymphoblast) from a single individual are stored in the Repository.

Famm #: Within each pedigree, each family member is assigned a unique family member number. Cultures having the same Famm# were obtained from the same individual.

Remarks: Includes any pertinent information that is not found in the preceding columns and cross references to other samples from the same individual and/or other members of the same family.

Special Notations:

* Before a Repository number indicates that a literature reference for this cell line is found in Appendix C, page 490.

➤ Before a Repository number indicates that a compliance form (page XVI) must be on file at the Repository before shipment of the culture can be made.

NIGMS Human Genetic Mutant Cell Repository

NIGMS
Human Genetic
Mutant Cell
Repository

Apparently Normal Human Fibroblasts

Appearance
Normal
Human
Fibroblasts

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Fetal Tissue										
GM0468	2	EEMEM	20U 0026	FW	M		PROBAND			46,XY; 14.5 cm crown-rump length; skin biopsy
*GM0011	2	EEMEM	20U 0026	8FW	M	BL	PROBAND			46,XY; skin biopsy; 3% of cells show random loss of one chromosome
*GM0010	2	EEMEM	20U 0026	12FW	M	BL	PROBAND	166	1	46,XY; skin biopsy; 12 cm crown-rump length; same patient as GM1380; 10% of cells show random loss of one chromosome and 2% show random gain of one chromosome
GM1380	13	EEMEM	20U 0026	12FW	M	BL	PROBAND	166	1	46,XY; lung biopsy; 12 cm crown-rump length; same patient as GM0010; 6% of cells show random loss/gain of one chromosome
*GM1379	4	EEMEM	20U 0026	12FW	M		PROBAND	167	1	46,XY; lung biopsy; 10.5 cm crown-rump length; same patient as GM1381; 15% of cells show random loss of one chromosome
*GM1381	4	EEMEM	20U 0026	12FW	M		PROBAND	167	1	46,XY; skin biopsy; 10.5 cm crown-rump length; same patient as GM1379; 12% of cells show random loss of one chromosome
*GM1603	2	EEMEM	20U 0152	12FW	M	BL	PROBAND	168	1	46,XY; skin biopsy; same patient as GM1604
*GM1604	2	EEMEM	20U 0152	12FW	M	BL	PROBAND	168	1	46,XY; lung biopsy; same patient as GM1603; 7% of cells show random loss of one chromosome
GM4522	3	EEMEM	10U 0331	16FW	F		PROBAND	739	1	Line SK118; skin biopsy (buttock); 46,XX
GM6111	4	EEMEM	20U 0331	16FW	F	BL	PROBAND	911	1	Skin biopsy; same patient as GM6112; 46,XX; 6% of cells show random chromosome loss
GM6112	3	EEMEM	20U 0331	16FW	F	BL	PROBAND	911	1	Lung biopsy; same patient as GM6111; 46,XX; 4% of cells show random chromosome loss
GM5386	1	EEMEM	20U 0152	20FW	M	WH	PROBAND	852	1	Skin biopsy; 14.0 cm crown-rump length; 46,XY; same patient as GM5387 & GM5421A
GM5387	1	EEMEM	20U 0152	20FW	M	WH	PROBAND	852	1	Lung biopsy; 14.0 cm crown-rump length; 46,XY; same patient as GM5386 & 5421A
GM5421A	5	EEMEM	20U 0152	20FW	M	WH	PROBAND	852	1	Conjunctiva biopsy; 14.0 cm crown-rump length; 46,XY; same patient as GM5386 & 5387

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Fetal Tissue										
GM5388	1	EEMEM	20U 0152	20FW	M	WH	TWIN	852	2	Skin biopsy; 13.5 cm crown-rump length; 46,XY; same patient as GM5389 & GM5420
GM5389	1	EEMEM	20U 0152	20FW	M	WH	TWIN	852	2	Lung biopsy; 13.5 cm crown-rump length; 46,XY; same patient as GM5388 & GM5420
GM5420	3	EEMEM	20U 0152	20FW	M	WH	TWIN	852	2	Conjunctiva biopsy; 13.5 cm crown-rump length; 46,XY; same patient as GM5388 & GM5389
GM6113	3	EEMEM	20U 0331	20FW	M	BL	PROBAND	910	1	Skin biopsy; same patient as GM6114; 46,XY; 10% of cells show random chromosome loss
GM6114	3	EEMEM	20U 0331	20FW	M	BL	PROBAND	910	1	Lung biopsy; same patient as GM6113; 46,XY; 12% of cells show random chromosome loss
GM6170	3	EEMEM	20U 0331	20FW	M	BL	PROBAND	915	1	Skin biopsy; 46,XY; 6% of cells show random chromosome loss; same patient as GM6171
GM6171	3	EEMEM	20U 0331	20FW	M	BL	PROBAND	915	1	Lung biopsy; 46,XY; 8% of cells show random chromosome loss/gain; same patient as GM6170
GM6168	4	EEMEM	20U 0331	20FW	M	BL	PROBAND	916	1	Skin biopsy; 46,XY; 8% of cells show random chromosome loss; same patient as GM6169
GM6169	4	EEMEM	20U 0331	20FW	M	BL	PROBAND	916	1	Lung biopsy; 46,XY; 6% of cells show random chromosome loss; same patient as GM6168
GM6166	5	EEMEM	20U 0331	20FW	M	BL	PROBAND	917	1	Skin biopsy; 46,XY; 6% of cells show random chromosome loss; same patient as GM6167
GM6167	3	EEMEM	20U 0331	20FW	M	BL	PROBAND	917	1	Lung biopsy; 46,XY; 2% of cells show random chromosome loss; same patient as GM6166
Apparently Normal: Non-Fetal Tissue										
*GM09708	9	EEMEM	20U 0095	3DA	M	WH	PROBAND			Skin biopsy; 46,XY
*GM3468	8	EEMEM	20U 0331	5DA	M	WH	PROBAND			Foreskin; sensitive to azaguanine and thioguanine; utilizes exogenous hypoxanthine; HPRT & APRT normal; 46,XY

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
*GM2936B	6	EEMEM	20U 0185	20DA	M	WH	PROBAND			Mexican; skin biopsy (leg); 46,XY; 8% of cells show random loss of one chromosome and 2% show random gain of one chromosome; 12% of cells show breakage in chromosomes #1, #2, & #6
GM5756	2	EEMEM	20U 0414	2MO	M	WH	PROBAND			Skin biopsy (inguinal); 46,XY; 6% of cells show random chromosome loss
*GM0041A	11	EEMEM	20U 0045	3MO	F	WH	DAU	162	1	46,XX; Puerto Rican; skin biopsy; mother is GM0037A; 3% of cells show random chromosome loss
*GM0302A	11	EEMEM	20U 0095	10MO	M		PROBAND	331	1	Skin biopsy; formerly GM0075; normal B-hexosaminidase and B-galactosidase; 46,XY; 12% of cells show random chromosome loss
GM5659	2	EEMEM	20U 0414	14MO	M	WH	PROBAND			Skin biopsy (chest); 46,XY
GM5758	2	EEMEM	20U 0414	14MO	M	BL	PROBAND			Skin biopsy (inguinal); 46,XY
*GM0969B	10	EEMEM	20U 0095	2	F	WH	PROBAND			46,XX; 10% of cells show random chromosome loss; skin biopsy
*GM0498A	7	EEMEM	20U 0128	3	M		PROBAND			Skin biopsy; 46,XY; 12% of cells show random chromosome loss
*GM2938C	8	EEMEM	20U 0185	3	M	WH	PROBAND			Skin biopsy (leg); 46,XY; 8% of cells show random chromosome loss
GM5565	2	EEMEM	20U 0414	3	M	WH	PROBAND			Hispanic; skin biopsy (inguinal); past history of asthma; 46,XY
*GM0497B	16	EEMEM	20U 0128	4	M	WH	PROBAND			Skin biopsy (forearm); 46,XY
*GM0408A	11	EEMEM	20U 0128	5	F	WH	PROBAND			46,XX; skin biopsy
GM5381	3	EEMEM	20U 0414	5	M	BL	PROBAND			Skin biopsy (umbilical); 46,XY; 6% of cells show random chromosome loss/gain; 6% are polyploid
*GM0409A		EEMEM	20U 0128	7	M	WH	PROBAND			46,XY; skin biopsy; 12% of cells show random chromosome loss; passage 8 at INR
GM5757	2	EEMEM	20U 0414	7	M	BL	PROBAND			Skin biopsy (inguinal); 46,XY
*GM0499A	11	EEMEM	20U 0128	8	M	WH	PROBAND			Skin biopsy; 46,XY

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FANM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
*GM0038A	8	EEMEM	20U 0045	9	F	BL	DAU	163	1	46,XX; 14% of cells show random chromosome loss; skin biopsy; mother is GM0043
*GM0500A	10	EEMEM	20U 0128	10	M	WH	PROBAND			Skin biopsy; 46,XY
*GM3348A	5	EEMEM	20U 0026	10	M	WH	PROBAND			Skin biopsy (inguinal); 46,XY
*GM3349	3	EEMEM	20U 0026	10	M	BL	PROBAND			Skin biopsy; 46,XY
GM1864A	6	EEMEM	20U 0125	11	M	WH	BRO	161	3	46,XY; normal RBC G6PD level; Xg(a) antigen absent; skin biopsy; brother of GM1863; 1 cell out of 60 showed chromosome structural aberration
*GM0323	4	EEMEM	20U 0125	11	M	WH	BRO	164	1	46,XY; skin biopsy (upper arm); Sardinian; normal RBC G6PD level, Xg(a) antigen positive; brother of GM0326(XXXXY); mother is GM0321 Fibroblast; 8% of cells show random chromosome loss
*GM1652	7	EEMEM	20U 0133	11	F	WH	PROBAND	165	1	46,XX; skin biopsy (left forearm); daughter of GM1650 and GM1653; granddaughter of GM1680 and GM1681; sister of GM1651A ; 8% of cells show random chromosome loss
GM2036	10	EEMEM	10U 0133	11	F	WH	SIS	203	2	Skin biopsy; sister of GM2037A; 46,XX; 8% of cells show random chromosome loss
*GM1582A	8	EEMEM	20U 0026	11	F	WH	PROBAND	348	1	Formerly GM0240; skin biopsy (left forearm); tendency to develop translocations in vitro; sister of GM2270A; 46,XX; 17% of cells show random chromosome loss; 3% show random gain of one chromosome
*GM0316A	9	EEMEM	20U 0026	12	M	WH	PROBAND			46,XY; skin biopsy (arm); 12% of cells show random chromosome loss
*GM1651A	11	EEMEM	20U 0133	13	F	WH	SIS	165	2	Skin biopsy (left forearm); daughter of GM1650 & GM1653; granddaughter of GM1680 & GM1681; sister of GM1652; 46,XX; 15% of cells show random chromosome loss
*GM2037A	10	EEMEM	10U 0133	13	M	WH	PROBAND	203	1	46,XY; skin biopsy; brother of GM2036; 6% of cells show random chromosome loss/gain
*GM0037A	8	EEMEM	20U 0045	18	F	WH	PROBAND	162	2	See GM0637B(SV40 transformed GM0037); skin biopsy; Puerto Rican; daughter is GM0041A; 46,XX

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
*GM2987A	6	EEMEM	20U 0185	19	M	WH	PROBAND			Skin biopsy (leg); 46,XY
*GM3377A	6	EEMEM	20U 0026	19	M	WH	PROBAND			46,XY; skin biopsy (forearm)
GM2270A	11	EEMEM	20U 0026	19	M	WH	BRO	348	2	Skin biopsy (forearm); brother of GM1582A; 46,XY; at P15, 4% of cells show structural aberrations; 2/50 cells are 46,X,t(X;16)
*GM3440	2	EEMEM	20U 0185	20	M	WH	PROBAND			Skin biopsy (leg); 46,XY
GM3523	3	EEMEM	20U 0026	21	M	WH	PROBAND			Skin biopsy (rt neck); 46,XY; 8% of cells show random chromosome loss
GM2937A	5	EEMEM	20U 0185	22	M	WH	PROBAND			46,XY; skin biopsy; 8% of cells examined show chromosome structural abnormalities
*GM5294	4	EEMEM	20U 0016	22	M	BL	PROBAND	844	1	Lymphocytes show trisomic codominant expression of HLA A (34,19,23) and B (13, 12,35) haplotypes; 46,XY in lymphocytes; in fibroblasts, 17% of cells have an additional t(3;11)(p21;q23) at P6
*GM3234	3	EEMEM	20U 0215	23	M	AI	PROBAND			Pima Indian; skin biopsy (medial forearm); 46,XY; 12% of cells show random chromosome abnormalities
GM4390	3	EEMEM	20U 0026	23	F	WH	PROBAND			Skin biopsy (left forearm); 46,XX
GM1891	6	EEMEM	20U 0125	24	M	WH	PROBAND			46,XY; normal RBC level of G6PD(B); skin biopsy; 10% of cells show random chromosome loss
*GM3652	3	EEMEM	20U 0026	24	M	WH	PROBAND			46,XY; skin biopsy (left forearm)
*GM3651C	5	EEMEM	20U 0026	25	F	WH	PROBAND			Skin biopsy (left forearm); 46,XX
*GM0726A	7	EEMEM	20U 0026	26	F	OR	PROBAND			Korean; skin biopsy (forearm); 46,XX; 8% of cells show random chromosome loss/gain
*GM2912A	4	EEMEM	20U 0185	26	M	WH	PROBAND			Skin biopsy; 46,XY; 6% of cells show random chromosome loss/gain
*GM2674	7	R1640	15U 0030	29	F	WH	PROBAND			Skin biopsy; 46,XX
*GM0495	6	EEMEM	20U 0045	29	M		PARENT	417	2	46,XY; formerly GM0104; skin biopsy (buttock); clinically normal parent of child with low growth hormone level; 15% of cells show random chromosome loss

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
							Apparently Normal: Non-Fetal Tissue			
*GM0023	5	EEMEM	20U 0095	31	F	WH	PARENT	492	1	46,XX; mother of Down's child; skin biopsy; wife of GM0024; 10% of fibroblasts show random chromosome loss
*GM0024	5	EEMEM	20U 0095	31	M	WH	PARENT	492	2	46,XY; father of Down's child; skin biopsy; husband of GM0023
*GM0043	5	EEMEM	20U 0045	32	F	BL	MOTHER	163	2	46,XX; skin biopsy; daughter is GM0038A
*GM0964B	7	EEMEM	20U 0026	33	M	WH	PROBAND			46,XY; skin biopsy (left forearm); 11% of cells show random chromosome loss
*GM2573	6	EEMEM	20U 0030	33	M	WH	PROBAND			Skin biopsy; 46,XY; large Y chromosome in all cells
*GM2185	6	EEMEM	20U 0186	36	M	WH	PROBAND	455	1	46,XY; normal spouse of Huntington's Disease patient; skin biopsy; see GM2184A Lymphoid; 5% of cells examined show chromosome structural aberrations
*GM1650	6	EEMEM	20U 0133	37	F	WH	MOTHER	165	3	Skin biopsy (left forearm); mother of GM1651A & GM1652; 46,XX; 4% of cells show chromosome structural aberrations
*GM1653	8	EEMEM	20U 0133	37	M	WH	FATHER	165	4	46,XY; skin biopsy (left forearm); father of GM1651A & GM1652; son of GM1680 and GM1681
GM2665	5	EEMEM	20U 0147	37	F	WH	PROBAND	524	1	Skin biopsy; slow growing culture; 46,XX; 26% of cells show random chromosome loss; 4% show random gain of one chromosome; 15% are polyploid; 12% show random chromosome abnormalities
GM1717	3	EEMEM	20U 0125	39	M	WH	PROBAND			46,XY; normal RBC G6PD level, Xg(a) antigen absent; skin biopsy (forearm)
GM0321	3	EEMEM	20U 0125	40	F	WH	MOTHER	164	2	46,XX; skin biopsy (upper arm); Sardinian; normal RBC G6PD level, Xg(a) antigen positive; sons are GM0323 and GM0326 (XXXXY)
*GM0275A	9	EEMEM	20U 0026	42	M	WH	PROBAND			46,XY; skin biopsy (upper rt arm); 4% of cells show random chromosome loss
*GM0730A	6	EEMEM	20U 0026	45	F	WH	PROBAND	343	1	46,XX; skin biopsy (forearm); formerly GM0238; 20% of cells show random chromosome loss

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
GM1863	3	EEMEM	20U 0125	46	M	WH	PROBAND	161	2	46,XY/46,XY,t(4;7)(4pter>4q21::7q36>7qter;7pter>7q36::4q21>4qter); 70%/30%; normal RBC G6PD level, Xg(a) antigen absent; skin biopsy; brother of GM1864A
GM5879	2	EEMEM	20U 0026	48	F	WH	PROBAND	887	1	Skin biopsy (left upper arm); see GM5920 Lymphoid; 46,XX; 6% of cells show random chromosome loss
*GM0967A	8	EEMEM	20U 0026	51	M	WH	PROBAND	476	1	46,XY; skin biopsy (left forearm); tendency to develop translocations in vitro; see GM1310B Lymphoid; 19% of cells show random chromosome loss/gain
*GM3527	5	EEMEM	20U 0026	55	M	WH	PROBAND			Skin biopsy (penis); 64% of fibroblasts show 46,XY,t(3;4)(3pter>3q22::4p14>4pter;4qter>4p14::3q22>3qter)
GM0967B	4	EEMEM	20U 0026	57	M	WH	PROBAND	476	1	Skin biopsy; 46,XY; tendency to develop translocations in vitro; same patient as GM0967A six years later; see GM1310A Lymphoid
GM4260	9	EEMEM	20U 0371	60	M	WH	PROBAND			46,XY; 14% of cells show random chromosome loss
*GM2623A	4	EEMEM	20U 0030	61	F	WH	PARENT			46,XX; clinically normal mother of an XX male
*GM0288A	10	EEMEM	20U 0035	64	M	WH	SPOUSE	288	2	Skin biopsy (rt forearm); formerly GM0095; 46,XY; wife is GM0305A (Huntington's disease fibroblast)
*GM3529	2	EEMEM	20U 0026	66	M	BL	PROBAND			Skin biopsy (left thigh); 46,XY
GM3524	4	EEMEM	20U 0026	67	F	BL	PROBAND			Skin biopsy (rt calf); 46,XX
GM3658	4	EEMEM	20U 0026	68	M	WH	PROBAND	542	1	Skin biopsy (left forearm); 46,XY: see GM3657 Lymphoid
*GM1681	8	EEMEM	20U 0133	70	M	WH	PAT GRFR	165	5	46,XY; skin biopsy (left forearm); father of GM1653; grandfather of GM1651A & GM1652; 6% of fibroblasts show random chromosome loss & 2% show random translocations
*GM1680	8	EEMEM	20U 0133	71	F	WH	PAT GRMR	165	6	46,XX; skin biopsy (left forearm); mother of GM1653; grandmother of GM1651A & GM1652; 8% of fibroblasts show random chromosome loss & 2% show random translocations

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
Apparently Normal: Non-Fetal Tissue										
GM3525	3	EEMEM 20U	0026	80	F	WH	PROBAND			Skin biopsy (abdomen); 46,XX; 12% of cells show random chromosome loss/gain
*GM1706A	9	EEMEM 20U	0026	82	F	WH	PROBAND	342	1	46,XX; formerly GM0237; skin biopsy (left forearm); 14% of cells show random chromosome loss
*GM0731A	8	EEMEM 20U	0135	96	M	WH	PROBAND			46,XY; skin biopsy

Biochemically Mutant Human Fibroblasts

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Alkaptonuria (Homogentisic Acid Oxidase Deficiency) - 20350												
GM2228	4	EEMEM 20U	0147	36	M	WH	--		PROBAND			Pedigree includes two affected brothers
Argininemia (Arginase Deficiency) - 20780												
*GM0954	5	EEMEM 20U	0006	4	F	WH	--		PROBAND	422	1	Mexican-American; red cell Arginase deficiency; see GM2011 Lymphoid
Argininosuccinicaciduria (Argininosuccinate Lyase Deficiency) - 20790												
*GM0525	8	EEMEM 20U	0123	9	M	WH	--	A	PROBAND	1	1	Argininosuccinase deficiency; similarly affected brother
*GM0533	8	EEMEM 20U	0123	20	M	WH	--	A	AFF BRO	1	2	Argininosuccinase deficiency
GM0540	8	EEMEM 20U	0123	63	M	WH	+-(0)	A	FATHER	1	3	Normal Argininosuccinic acid lyase activity; two affected sons
*GM2830	7	EEMEM 20U	0270	8	F	WH	--	A	PROBAND	325	1	Argininosuccinase deficiency
GM2831	10	EEMEM 20U	0270	38	F	WH	+-(0)		MOTHER	325	2	Clinically unaffected
GM2832	7	EEMEM 20U	0270	48	M	WH	+-(0)		FATHER	325	3	Clinically unaffected
Citrullinuria (Citrullinemia) - 21570												
*GM1684	6	EEMEM 20U	0114	1DA	F	WH	--	A	PROBAND			Neonatal; deficient Argininosuccinate synthetase; no detectable enzyme antigen
*GM4326	5	EEMEM 20U	0266	2	F	WH	--	B	PROBAND			No detectable Argininosuccinate synthetase activity; growth and developmental delay; parents are first cousins
*GM1044	3	EEMEM 20U	0107	8MO	M	WH	--	A	PROBAND	2	1	See GM1204 Lymphoid; deficient Argininosuccinate synthetase; no detectable enzyme antigen; no differences observed for control and patient mRNA using S1 nuclease mapping
GM1058	3	EEMEM 20U	0107	27	M	WH	+-(0)	A	FATHER	2	2	See GM1206 Lymphoid; 38% of normal Argininosuccinate synthetase activity in fibroblasts
*GM1679	2	EEMEM 20U	0157	1DA	M	WH	--	A	PROBAND	3	1	Mild mental retardation; increased plasma citrulline; a sib died of citrullinemia; deficient ASS activity; no detectable enzyme antigen; possible compound heterozygote by mRNA analysis; same pt. as GM3056; see GM1685 Lymphoid
*GM3056	6	EEMEM 20U	0134	3	M	WH	--	A	PROBAND	3	1	Mild mental retardation; increased plasma citrulline; a sib died of citrullinemia; deficient ASS activity; no detectable enzyme antigen; possible compound heterozygote by mRNA analysis; same pt. as GM1679; see GM1685 Lymphoid

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
GM0046	5	EEMEM 20U	0121	3	M	WH	--	A	PROBAND			Increased intracellular cystine in fibroblasts
GM0706	3	EEMEM 20U	0117	18MO	M	WH	--	•	PROBAND			Pedigree includes an affected sib
GM2066	2	EEMEM 20U	0019	1	F	WH	--	A	PROBAND			Increased intracellular cystine in fibroblasts
*GM0090A	6	EEMEM 20U	0114	24FW	M	WH	--	A	PROBAND	4	1	Lung fibroblasts; positive family history; increased intracellular cystine in fibroblasts; same patient as GM0304
*GM0304	4	EEMEM 20U	0114	24FW	M	WH	--	A	PROBAND	4	1	Skin fibroblasts; formerly GM0091; increased intracellular cystine in fibroblasts; same patient as GM0090A
*GM0489	10	EEMEM 10U	0114	18MO	M	WH	--	A	AFF BRO	4	2	Fibroblasts show increased intracellular cystine
GM0760	2	EEMEM 20U	0128	24FW	M	WH	--	A	PROBAND	5	1	Fibroblasts also available from bone marrow, kidney, thymus, liver, thyroid, and amnion; slow growing culture; increased intracellular cystine in fibroblasts
GM0018	4	EEMEM 20U	0121	3	M	WH	--	A	PROBAND	248	1	Increased intracellular cystine in fibroblasts; similarly affected brother
GM0020	4	EEMEM 20U	0121	5MO	M	WH	--	A	AFF BRO	248	2	Increased intracellular cystine in fibroblasts
*GM0008	5	EEMEM 20U	0121	5	F	WH	--	A	PROBAND	478	1	Increased intracellular cystine in fibroblasts
GM6966	6	EEMEM 20U	0114	6	M	WH	--		PROBAND	978	1	Cystic fibrosis and clinically documented nephropathic cystinosis; growth difficulty; recurrent pneumonias; hyponatremia & other electrolyte abnormalities; renal disease; cystine crystals in cornea
Cystinosis: Benign Type; Adult Type - 21975												
GM0378	8	EEMEM 10U	0114	7	F	WH	--		PROBAND	6	1	Conjunctiva and cornea contain typical cystinotic crystals; similarly affected brother
GM0379	8	EEMEM 10U	0114	4	M	WH	--		AFF BRO	6	2	Conjunctiva and cornea contain typical cystinotic crystals

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DISORDERS OF AMINO ACID METABOLISM												
Cystinosis: Benign Type; Adult Type - 21975												
,GM0906	11	EEMEM 20U	0114	32	M	WH	+-(O)		FATHER	6	3	Two affected children
GM0907	15	EEMEM 20U	0114	30	F	WH	+-(O)		MOTHER	6	4	Two affected children
Cystinosis: Late Onset Juvenile or Adolescent Type - 21990												
GM0908	8	EEMEM 20U	0114	12	M	WH	--	B	PROBAND	7	1	Nephropathic cystinosis; fibroblasts show increased levels of intracellular cystine
GM0909	9	EEMEM 20U	0114	42	F	WH	+-(O)		MOTHER	7	2	Clinically unaffected
GM0910A	11	EEMEM 20U	0114	49	M	WH	+-(O)		FATHER	7	3	Clinically unaffected
Glutaricacidemia; Glutaricaciduria Type I - 23167												
*GM5002	6	EEMEM 20U	0123	2	M	WH	--	B	PROBAND			Delayed motor & mental development, dystonia, spasticity, choreoathetosis, & metabolic acidosis; deficient Glutaryl-CoA dehydrogenase activity; negative family history
Glutaricaciduria IIB; Ethylmalonic-Adipicaciduria - 23168												
GM3131		EEMEM 20U	0302	19	F		--		PROBAND			Hypoglycemia and organic aciduria; two similarly affected sibs; passage 2 at IMR
Gyrate Atrophy of Choroid and Retina (Ornithinemia) - 25887												
GM6330	3	EEMEM 20U	0319	27	F	WH	--	A	PROBAND			Increas. serum & urine ornithine levels; progressive blindness; no sign of neurologic involvement; severe constrict. of visual fields; flat ERG; neg family history; <5% of norm. ornithine aminotransferase act.; unrespon in vitro to Vit B6
GM6335	2	EEMEM 20U	0319	12	M	WH	--	A	PROBAND	953	1	Elevated serum & urine ornithine levels; loss of vis acuity; flat ERG; discrete spinocerebel ataxia; norm musc morphology; sim aff sib; parents 2nd cousins; < 5 % of normal ornithine aminotransferase act; unrespon in vitro to Vitamin B6
GM6336	2	EEMEM 20U	0319	22	F	WH	--	A	AFF SIS	953	2	Progress. blindness since 12; flat ERG; spinocerebel atax.; atrophy of leg & ft. mus; abnor musc bx.; elevat. serum ornithine level; sim aff sib; parents 2nd cous.; < 5% normal ornithine aminotransferase act.; unrespon in vitro to Vit B6
Hartnup Disease - 23450												
*GM5003	8	EEMEM 20U	0123	18	M	WH	--		PROBAND	814	1	Mental retardation & typical aminoaciduria; defective intestinal transport of tryptophan & phenylalanine; similarly affected brother

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DISORDERS OF AMINO ACID METABOLISM												
Hartnup Disease - 23450												
*GM5004	6	EEMEM 20U	0123	15	M	WH	--		AFF BRO	814	2	Mental retardation & typical aminoaciduria; similarly affected brother; defective intestinal transport of tryptophan & phenylalanine
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
*GM0342	3	EEMEM 20U	0066	20	F	WH	--	A	PROBAND			Excessive urinary homocystine; low IQ; bone pain; seizures; ectopic lens; osteoporosis; spinal compression fractures; B6 responsive; deficient cystathionine synthetase activity
GM0423	8	EEMEM 20U	0114		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0424A	12	EEMEM 20U	0114		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0625	3	EEMEM 20U	0038	35	M	WH	+-(0)	A	PARENT			Confirmed heterozygote; two affected children
GM0720	3	EEMEM 20U	0038	30	M	WH	--	B	PROBAND			Def. cystathionine synthetase activity
GM0724	14	EEMEM 20U	0038		M	WH	--		PROBAND			Clinically affected
GM0664	12	EEMEM 20U	0038	13	F		--	A	PROBAND			Def. cystathionine synthetase activity
GM0865	5	EEMEM 20U	0038		M		--	A	PROBAND			Def. cystathionine synthetase activity
GM0584	3	EEMEM 20U	0038	39	M	WH	--	A	PROBAND	8	1	Def. cystathionine synthetase activity
GM0585	3	EEMEM 20U	0038	65	M	WH	+-(0)		FATHER	8	2	Clinically unaffected
GM0594	2	EEMEM 20U	0086	23	M	WH	--	A	PROBAND	9	1	Def. cystathionine synthetase activity
GM0417	4	EEMEM 20U	0042	42	F	WH	+-(0)		MOTHER	9	2	Clinically unaffected
GM0722	7	EEMEM 20U	0038	24	M	WH	--	B	PROBAND	10	1	Clinically atypical; formerly GM0548
GM0725	7	EEMEM 20U	0038		F	WH	+-(0)		MOTHER	10	2	Clinically unaffected
GM0751	3	EEMEM 20U	0086	19	M	WH	--	A	PROBAND	11	1	Def. cystathionine synthetase activity; two similarly affected brothers
GM0752	3	EEMEM 20U	0086	23	M	WH	--	A	AFF BRO	11	2	Def. cystathionine synthetase activity; two similarly affected brothers
GM0753A	5	EEMEM 20U	0086	41	M	WH	+-(0)	A	FATHER	11	4	Confirmed heterozygote; three affected children

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DISORDERS OF AMINO ACID METABOLISM												
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
GM0883	2	EEMEM 20U	0086	42	F	WH	+-(0)		MOTHER	11	5	Three affected children; clinically unaffected
GM0885	3	EEMEM 20U	0086	17	M	WH	--	B	AFF BRO	11	6	Def. cystathionine synthetase activity; two similarly affected brothers
GM0813	2	EEMEM 20U	0038	13	M	WH	+-	A	MAT 1ST COU	11	7	Confirmed heterozygote
GM0866	8	EEMEM 20U	0038	10	F		--	A	PROBAND	12	1	Def. cystathionine synthetase activity; similarly affected sib
GM0867	10	EEMEM 20U	0038	6	M		--	A	AFF BRO	12	2	Def. cystathionine synthetase activity
GM1128	3	EEMEM 20U	0038	11	F	WH	--	A	PROBAND	14	1	B6 non-responsive; def. cystathionine synthetase activity; similarly affected sib
GM1129	3	EEMEM 20U	0038	9	M	WH	--	B	AFF BRO	14	2	B6 non-responsive
GM1126	3	EEMEM 20U	0038		M	WH	+-(0)		FATHER	14	3	Two affected children; clinically unaffected
GM1374	4	EEMEM 20U	0038	13	F	WH	--	A	PROBAND	15	1	Def. cystathionine synthetase activity
GM1375	6	EEMEM 20U	0038		F	WH	+-(0)	A	MOTHER	15	2	Confirmed heterozygote; clinically unaffected
GM1376	6	EEMEM 20U	0038		M	WH	+-(0)	A	FATHER	15	3	Confirmed heterozygote; clinically unaffected
GM0721	7	EEMEM 20U	0038	11	F	WH	--	B	PROBAND	639	1	Def. cystathionine synthetase activity; formerly GM0547
Ornithine Transcarbamylase Deficiency: Hyperammonemia I - 31125												
*GM6902	4	EEMEM 20U	0006	6	M	WH	Y-		PROBAND			Positive family history for dietary protein intolerance by the mother and neonatal death in the maternal uncle; minimal awareness of surroundings; hypertonic; no detectable liver OTC activity in liver biopsy
Carbamoyl Phosphate Synthetase I Deficiency: Hyperammonemia II - 23730												
GM3432	8	EEMEM 20U	0049	4MO	F	WH	--		PROBAND			Canadian Rep. No. 615; 46,XX; lethal neonatal hyperammonemia; Pakistani; CPS deficiency confirmed by liver biopsy; positive family history; parents are cousins

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM											
Carbamoyl Phosphate Synthetase I Deficiency: Hyperammonemia II - 23730											
*GM4311	5	EEMEM 20U	0266	18	F	WH	--	PROBAND			Onset of lethargy and vomiting at age 3 weeks; severe mental retardation; growth retardation; CPS deficiency confirmed by liver biopsy
Hyperglycinemia: Non-Ketotic - 23830											
GM0747	3	EEMEM 20U	0107	18MO	F	WH	--	PROBAND			Clinically affected
GM1297	8	EEMEM 20U	0114		M		--	PROBAND			Clinically affected
GM1301	8	EEMEM 20U	0114	2MO	F	WH	--	PROBAND			Clinically affected
GM1302	9	EEMEM 20U	0114	1MO	F		--	PROBAND			Clinically affected
GM0880	5	EEMEM 20U	0095	21	M	WH	--	PROBAND	16	1	Spastic paraparesis; two affected sibs
GM1140		EEMEM 20U	0095	16	M	WH	--	AFF BRO	16	2	Passage 3 at IMR; two affected sibs
GM1162		EEMEM 20U	0095	28	M	WH	--	AFF BRO	16	3	Passage 3 at IMR; two affected sibs
Hyperlysinemia: Lysine-Ketoglutarate Reductase Deficiency - 23870											
GM7280	5	EEMEM 10U	0496	22	M	WH	-- B	PROBAND			Spastic diplegia with lower extremity atrophy; mild mental deficit; occasional vomiting; avoids protein foods; confined to a wheelchair; elevated plasma & urine lysine; 7-8% of normal lysine catabolism in fibroblasts
Hypermethioninemia - 23891											
GM0911	2	EEMEM 20U	0086	3MO	M	WH	--	PROBAND			Deficient hepatic methionine adenosyltransferase activity; normal activity in fibroblasts
Hyperpipecolatemia - 23940											
*GM3605	7	EEMEM 20U	0134	7	M	WH	--	PROBAND			Hepatomegaly; severe mental retardation; developmental delay; functional blindness; elevated pipecolic acid in serum
Hyperphenylalaninemia - 26158											
GM0006	7	EEMEM 20U	0095	5	F	WH	--	PROBAND	17	1	Normal level of dihydropteridine reductase in skin fibroblasts; 46,XX
GM0004	9	EEMEM 20U	0095	33	M	WH	+-(O)	FATHER	17	2	46,XY; clinically unaffected
GM0007	7	EEMEM 20U	0095	30	F	WH	+-(O)	MOTHER	17	3	46,XX; normal level of dihydropteridine reductase in skin fibroblasts; clinically unaffected

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DISORDERS OF AMINO ACID METABOLISM											
Isovalericacidemia (Isovaleric Acid CoA Dehydrogenase Deficiency) - 24350											
GM0947	5	EEMEM 20U	0002	13	M	WH	--	B PROBAND			Clinically affected; fibroblasts are deficient in ability to decarboxylate leucine
GM6836	7	EEMEM 20U	0464	2MO	F	OT	--	PROBAND			BTack/Latin American; acidosis with increased serum isovaleric acid; seizures; neuromuscular irritability; profound hypocalcemia; elevated serum ammonia and hepatomegaly; similarly affected sib
*GM7490	5	EEMEM 20U	0505	3	M	BL	--	PROBAND			Intermittent vomiting and lethargy followed by transient alopecia and pancytopenia; no foul odor; urine organic acid analysis showed excessive isovalerylglycine; developmental delay; physical weakness; negative family history
Maple Syrup Urine Disease (Branched-Chain Ketoaciduria) - 24860											
*GM0296	15	EEMEM 20U	0033	5	M	BL	--	B PROBAND			Quadriplegic and severely retarded; a brother also has MSUD; 2-5% of normal branched-chain amino acid decarboxylase activity
*GM0297	18	EEMEM 20U	0033	13DA	F	WH	--	A PROBAND			Deficient (1%) branched-chain keto-acid decarboxylase activity; high urinary leucine, isoleucine, & valine, and their branched-chain alpha-keto acid derivatives
*GM0612	2	EEMEM 20U	0074	2MO	F	WH	--	A PROBAND			Less than 2% of normal branched-chain keto-acid decarboxylase activity
GM1744	11	EEMEM 20U	0077	13DA	F	WH	--	A PROBAND			Deficient branched-chain keto-acid decarboxylase activity
GM1938	5	EEMEM 20U	0077	5	M	WH	--	A PROBAND			Deficient branched-chain keto-acid decarboxylase activity
GM5074	1	EEMEM 20U	0401	21DA	M	WH	--	PROBAND			Similarly affected sib; typical urine odor; severe muscle hypotonia; absent neonatal reflexes; high levels of valine, isoleucine, & leucine in plasma; expired at age 1 mo.
*GM0649	3	EEMEM 20U	0038	9MO	M	WH	--	A PROBAND	18	1	Elevated urine leucine, isoleucine, and valine; delayed psychomotor development; abnormal EEG; deficient branched-chain alpha-keto acid decarboxylase activity

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Maple Syrup Urine Disease (Branched-Chain Ketoaciduria) - 24860												
*GM0650	3	EEMEM 20U	0038		M	WH	+-(O)	A	FATHER	18	2	30-60% of normal branched-chain alpha-keto acid decarboxylase activity
*GM0651	3	EEMEM 20U	0038		F	WH	+-(O)	A	MOTHER	18	3	30-60% of normal branched-chain alpha-keto acid decarboxylase activity
GM1364	2	EEMEM 20U	0107	7	F	BL	--	B	PROBAND	20	1	See GM1366 Lymphoid; classical symptoms; 1 previously affected male sib
GM1654	2	EEMEM 20U	0107	5	F	WH	--	A	PROBAND	21	1	See GM1655 Lymphoid; two female sibs died of MSUD; Mennonite kindred; def. branched-chain keto-acid decarboxylase
GM1099	3	EEMEM 20U	0107	8MO	F	WH	--		PROBAND	266	1	Mennonite; hypotonia; seizures; positive family history
GM2327	3	EEMEM 20U	0107	4MO	F	WH	--	A	AFF SIS	266	2	Mennonite; pericardium biopsy; deficient branched-chain keto-acid decarboxylase
GM0016	4	EEMEM 20U	0095	25	M	WH	+-(O)	A	PARENT	491	1	Father of an affected child; proband not in Repository; deficient branched-chain keto-acid decarboxylase activity; 46+XY; spouse of GM0022; father of GM0242
GM0022	5	EEMEM 20U	0095	25	F	WH	+-(O)	A	PARENT	491	2	Mother of an affected child; proband not in Repository; deficient branched-chain keto-acid decarboxylase activity; 46+XX; spouse of GM0016; mother of GM0242
GM0242	4	EEMEM 20U	0095	1WK	F	WH	++	A	SIB	491	3	Normal branched-chain keto-acid decarboxylase activity; 46+XX; 6% of cells show random chromosome loss; parents are GM0016 & GM0022
Beta-Methylcrotonylglycinuria Types I and II - 21020 & 21021												
*GM3522	2	EEMEM 20U	0128	4MO	M	WH	--	A	PROBAND			Type II; holocarboxylase synthetase def; acidosis; "cat urine" odor; shock; responded well to biotin treatment; 1 previously affected sib; deficient propionyl CoA carboxylase, pyruvate carboxylase, & B-methylcrotonyl CoA carboxylase
Methylmalonicaciduria Types I & II - 25100, 25110, 25111												
*GM0050	13	EEMEM 20U	0095	1	M	WH	--	A	PROBAND			Unresponsive to B12; ATCC CCL 124; defective methylmalonyl CoA mutase apoenzyme; G6PD type B
GM0212	6	EEMEM 20U	0095	1	M	WH	--	A	PROBAND			Responsive to B12; cobalamin A mutation

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DISORDERS OF AMINO ACID METABOLISM												
Methylmalonicaciduria Types I & II - 25100, 25110, 25111												
GM0876	5	R1640 20U	0010	1MO	M	WH	--	A	PROBAND			Cobalamin B mutant; B12 responsive
GM0930		EEMEM 20U	0032	2	F	WH	--	A	PROBAND			Methylmalonyl CoA mutase defect; unresponsive to B12; passage 3 at IMR
GM1673	8	EEMEM 10U	0088	1DA	M	WH	--	B	PROBAND			Apomutase mutant
*GM1674	7	EEMEM 20U	0088	1DA	F	WH	--	B	PROBAND			Cobalamin A mutant; line no. 245
*GM0595	2	EEMEM 20U	0088	7	M	WH	--		PROBAND	22	1	Cobalamin A mutant; B12 responsive
GM0596	2	EEMEM 20U	0088	25	F	WH	+-(D)		MOTHER	22	2	Clinically unaffected
GM0306	6	EEMEM 20U	0095	2	M		--	A	PROBAND	315	1	Responsive to B12; cobalamin A mutation; formerly GM0051
Methylmalonicaciduria with Homocystinuria - 27740, 27741												
*GM2452	10	EEMEM 10U	0218	1	M	WH	--	B	PROBAND			Cbl D complementation group; normal methylmalonyl-CoA mutase activity and crossreacting material with anti-mutase antiserum
GM2453	17	EEMEM 10U	0218	7	F	WH	--	B	PROBAND			Cbl C complementation group
Phenylketonuria (Phenylalanine Hydroxylase Deficiency) - 26160												
GM0937	2	EEMEM 20U	0107	19MO	M	WH	--		PROBAND			Normal level of dihydropteridine reductase in skin fibroblasts; clinically affected
GM2406	2	EEMEM 20U	0074	2MO	M	WH	--		PROBAND			Pyloric stenosis; normal level of dihydropteridine reductase in skin fibroblasts; clinically affected
Propionicacidemia - (Hyperglycinemia with Ketoacidosis & Leukopenia) - 23200												
GM0056A	14	EEMEM 20U	0088	1	F	WH	--	B	PROBAND			Deficient Propionyl CoA carboxylase activity
GM0057	10	EEMEM 20U	0088	6	F	WH	--	B	PROBAND			Deficient Propionyl CoA carboxylase activity
GM1298	8	EEMEM 20U	0114	4	M		--	B	PROBAND			Deficient Propionyl CoA carboxylase activity
GM1299	7	EEMEM 20U	0114	2	M		--	B	PROBAND			Deficient Propionyl CoA carboxylase activity
GM1300	8	EEMEM 20U	0114	30MO	F		--	B	PROBAND			Deficient Propionyl CoA carboxylase activity

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DISORDERS OF AMINO ACID METABOLISM												
Propionicacidemia - (Hyperglycinemia with Ketoacidosis & Leukopenia) - 23200												
GM2805	8	EEMEM 20U	0070	1	M	WH	--		PROBAND			Hyperglycinuria
GM3590	7	EEMEM 20U	0088	22FW	M	WH	--	B	PROBAND			Deficient Propionyl CoA carboxylase activity; complementaion group C
GM0371	3	EEMEM 20U	0105	7WK	F	WH	--	B	PROBAND	23	1	Deficient Propionyl CoA carboxylase activity
GM0403	7	EEMEM 20U	0105	24	F	WH	+-(O)		MOTHER	23	2	Clinically unaffected
GM0405	7	EEMEM 20U	0105	25	M	WH	+-(O)		FATHER	23	3	Clinically unaffected
Pyroglutamicaciduria: 5-oxoprolinuria - 26613												
*GM3877	4	EEMEM 20U	0117	3	M	WH	--		PROBAND	569	1	Metabolic acidosis; hemolytic anemia; similarly affected sib
*GM3878	3	EEMEM 20U	0117	5	M	WH	--		AFF BRO	569	2	Metabolic acidosis; hemolytic anemia; fibroblasts deficient in glutathione synthetase activity
Tyrosine Aminotransferase Deficiency & Richner-Hanhart Syndrome - 27660												
*GM4374	12	EEMEM 20U	0267	5	F	AI	--	B	PROBAND			Seizures; mental retardation; normal fibroblast p-hydroxyphenylpyruvic hydroxylase activity; deficient cytosolic tyrosine aminotransferase activity
Tyrosinemia - 27670												
GM0286	7	EEMEM 20U	0095	2MO	M		--		PROBAND	319	1	Formerly GM0058; clinically affected
DISORDERS OF CARBOHYDRATE METABOLISM												
Amyloidosis I (Andrade Type; Portuguese Type of Amyloid Neuropathy) - 10480												
*GM1998	3	EEMEM 20U	0155	16	F	WH	+-		PROBAND	421	1	Non-Portuguese; see GM1999 Lymphoid; 4 affected generations (all female); vitreous opacities & perivascular exudates; affected mother died at age 21
Aspartylglycosaminuria (N-Aspartyl-Beta-Glucosaminidase Deficiency) - 20840												
GM0568	2	EEMEM 20U	0004	18	F	WH	--	A	PROBAND			Finnish; less than 10% of normal N-aspartyl-beta-glycosaminidase activity
*GM2056	3	EEMEM 20U	0004	32	M	WH	--	A	PROBAND			Finnish; less than 10% of normal N-aspartyl-beta-glycosaminidase activity
*GM2057	4	EEMEM 20U	0004	27	F	WH	--	A	PROBAND			Finnish; less than 10% of normal N-aspartyl-beta-glycosaminidase activity
GM3560	8	EEMEM 20U	0134	10	M	BL	--	A	PROBAND			Deficient N-aspartyl-beta-glycosaminidase; mental retardation and dysmorphic features

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Fructose-1,6-Diphosphatase Deficiency - 22970												
GM0282	6	EEMEM 20U	0095	7	F		--		PROBAND	322	1	Formerly GM0061; clinically affected
Fucosidosis - 23000												
GM0801	6	EEMEM 20U	0138	15	M	WH	--	B	PROBAND			Type II; Spanish-American; alpha-L-fucosidase deficient
*GM0802	5	EEMEM 20U	0138	7	M	WH	--	A	PROBAND			Type II; Spanish-American; alpha-L-fucosidase deficient; positive family history
*GM1214	5	EEMEM 20U	0108	18	M	WH	--		PROBAND			Type II; positive family history; alpha-L-fucosidase deficiency in serum and leukocytes
*GM0292	3	EEMEM 20U	0078	4	M	WH	--	A	PROBAND	24	1	Type II; deficient alpha-L-fucosidase; 46+XY; similarly affected sib; see GM1025 Lymphoid
*GM0291	3	EEMEM 20U	0078	9	M	WH	--	A	AFF BRO	24	2	Type II; deficient alpha-L-fucosidase; see GM1024 Lymphoid
*GM0290	3	EEMEM 20U	0078	29	F	WH	+-(0)	A	MOTHER	24	3	See GM1026 Lymphoid; low-normal alpha-L-fucosidase activity; two affected children
*GM0289	3	EEMEM 20U	0078	31	M	WH	+-(0)	A	FATHER	24	4	See GM1023 Lymphoid; low-normal alpha-L-fucosidase activity; two affected children
Galactosemia (Kinase Deficiency) - 23020												
*GM0334A		EEMEM 20U	0095	22	F	WH	--	A	PROBAND	25	1	Cataracts; deficient Galactokinase activity; passage 10 at IMR; slow growing culture
*GM0335		EEMEM 20U	0095	53	F	WH	+-(0)	A	MOTHER	25	2	Passage 3 at IMR; clinically unaffected; 58% of control galactokinase activity in fibroblasts
*GM0336		EEMEM 20U	0095	62	M	WH	+-(0)	A	FATHER	25	3	Passage 4 at IMR; clinically unaffected; 79% of control galactokinase activity in fibroblasts
Galactosemia (Transferase Deficiency) - 23040												
GM0422		EEMEM 20U	0095	9	F	WH	--		PROBAND			Passage 3 at IMR; clinically affected
GM0528		EEMEM 20U	0095	2	F	WH	--		PROBAND			Passage 4 at IMR; clinically affected
GM0727A	8	EEMEM 20U	0095	2MO	F	WH	--		PROBAND			No detectable transferase activity in erythrocytes

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Galactosemia (Transferase Deficiency) - 23040												
*GM1996	6	EEMEM 10U	0002	1	M	WH	+-	A	PROBAND			Duarte variant-galactosemia heterozygote; reduced transferase activity in fibroblasts and erythrocytes
*GM0052	11	EEMEM 10U	0095	8	M	BL	--	B	PROBAND	26	1	G6PD type B; ATCC CCL 133; see also GM0639 (SV40 transformed) and GM2796 Lymphoid; no detectable transferase activity in RBCs or fibroblasts; similarly affected brother; same patient as GM1908
GM1908	2	EEMEM 20U	0095	15	M	BL	--	B	PROBAND	26	1	Same patient as GM0052; earlier passage number; see also GM0639 (SV40 transformed) and GM2796 Lymphoid; no detectable transferase activity in RBCs or fibroblasts; similarly affected brother
*GM0053	12	EEMEM 10U	0095	6	M	BL	--	B	AFF BRO	26	2	G6PD Type A; ATCC CCL 132; no detectable transferase activity in RBCs or fibroblasts
*GM0054	10	EEMEM 20U	0095	3MO	M	WH	--	B	PROBAND	27	1	See also GM0638 (SV40 Transformed) and GM2795 Lymphoid; G6PD type B; same patient as GM1907; deficient transferase & normal kinase activity in fibroblasts
GM1907	3	EEMEM 20U	0095	6	M	WH	--	A	PROBAND	27	1	Same patient as GM0054; earlier passage number; deficient transferase and normal kinase activity in fibroblasts; see also GM0638 (SV40 transformed) and GM2795 Lymphoid
*GM0441	6	EEMEM 20U	0069	15	M	WH	--	A	PROBAND	28	1	See GM2412B Lymphoid; no detectable transferase activity; 46,XY; similarly affected sister
*GM0440	6	EEMEM 20U	0069	16	F	WH	--	A	AFF SIS	28	2	See GM1028 Lymphoid; no detectable transferase activity; 46,XX/46,X,t(X;11) - balanced; 92%/8%
*GM0439	6	EEMEM 20U	0069	41	F	WH	+-(O)	B	MOTHER	28	3	See GM1029 Lymphoid; 50% of normal transferase activity in erythrocytes; 2 affected children; 46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter) - balanced
*GM0438	6	EEMEM 20U	0069	46	M	WH	+-(O)	B	FATHER	28	4	50% of normal transferase activity in erythrocytes; 2 affected children; 46,XY

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DISORDERS OF CARBOHYDRATE METABOLISM												
Galactosemia (Transferase Deficiency) - 23040												
*GM0442	6	EEMEM 20U	0069	12	M	WH	++	B	BRO	28	5	Normal transferase activity in erythrocytes; 46,XY; 2 affected sibs
*GM1533	7	EEMEM 20U	0069	19	F	WH	++	B	SIS	28	6	Normal transferase and kinase activity in fibroblasts; 46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter)mat; balanced; normal X is late replicating; 2 affected sibs
GM1209	2	EEMEM 20U	0045	9MO	M	WH	--	A	PROBAND	29	1	Identical twin (single placenta) of GM1210; no detectable transferase activity
GM1210	2	EEMEM 20U	0045	9MO	M	WH	--	A	AFF TWIN	29	2	Identical twin (single placenta) of GM1209; no detectable transferase activity
GM1211	3	EEMEM 20U	0045	4	M	WH	+-	A	BRO	29	3	50% of normal transferase activity; clinically unaffected; 2 affected sibs
GM1212	3	EEMEM 20U	0045	29	F	WH	+-(O)	A	MOTHER	29	4	50% of normal transferase activity; 2 affected children (twins)
GM1208	3	EEMEM 20U	0045	27	M	WH	+-(O)		FATHER	29	5	Two affected children (twins)
GM1417	6	EEMEM 20U	0095	22	F		--		PROBAND	30	1	Slow growing culture; no detectable transferase activity in erythrocytes
GM1418	5	EEMEM 20U	0095		F		+-(O)		MOTHER	30	2	Slow growing culture; carrier level of transferase in erythrocytes
GM1419	5	EEMEM 20U	0095		M		+-(O)		FATHER	30	3	Slow growing culture; carrier level of transferase in erythrocytes
GM1703	4	EEMEM 20U	0095	9MO	M	WH	--	B	PROBAND	31	1	2nd generation with transferase deficiency Galactosemia
GM1704	3	EEMEM 20U	0095	2	M	WH	--	B	AFF COU	31	2	2nd generation with transferase deficiency Galactosemia
GM1741	8	EEMEM 20U	0077	24FW	M	WH	--	B	PROBAND	32	1	See GM1743 Amniotic; clinically affected
*GM0433	3	EEMEM 20U	0095	10	F	BL	--	A	PROBAND	300	1	Fibroblasts accumulate large amounts of intracellular gal-1-phosphate and galactitol
Glycogen Storage Disease: Type I - Von Gierke Disease - 23220												
GM0574	8	EEMEM 20U	0130	25	M	WH	--		PROBAND			Absent hepatic glucose-6-phosphatase

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM											
Glycogen Storage Disease: Type 1B - G6P Transport Defect - 23222											
*GM3719	9	EEMEM 10U	0006	5	M	OT	--	PROBAND			Type Ib; Panamanian Ind.; severe hypoglycemia, lactic acidosis, & neutropenia; liver bx shows G6P transport defect
Glycogen Storage Disease: Type II - Pompe Disease - 23230											
*GM0244		EEMEM 20U	0002	5MO	F	WH	--	A PROBAND			Mexican; deficient acid-alpha-1,4 glucosidase activity; CRM negative; 46,XX; passage 4 at IMR
*GM0248		EEMEM 20U	0095	5MO	M	BL	--	A PROBAND			Deficient acid-alpha-1,4 glucosidase; CRM negative; passage 3 at IMR
*GM0338	2	EEMEM 20U	0008	1MO	M	BL	--	A PROBAND			Respiratory distress, cardiomegaly, and enlarged liver; a sib died at age 3 mos. with cardiomegaly; absent acid-alpha-1,4 glucosidase activity; CRM negative; culture grows poorly
*GM0443		EEMEM 20U	0099	30	M		--	A PROBAND			Adult form; onset in third decade; 9 to 23% of normal acid-alpha-1,4 glucosidase activity; CRM positive; passage 3 at IMR
*GM1935	3	EEMEM 20U	0126	30	F	BL	--	A PROBAND			Adult form; onset of generalized muscle wasting & weakness at age 30; history of amenorrhea & hypertension; 1 to 3% of normal acid-alpha-1,4 glucosidase activity; CRM negative
GM3329	2	EEMEM 20U	0107	23MO	M	BL	--	A PROBAND			Deficient acid-alpha-1,4 glucosidase
*GM4271	32	EEMEM 20U	0193	2	M	WH	--	A PROBAND			"Muscular variant"; progressive amyotonic syndrome; glycogen storage disease of skeletal muscles; deficient acid-alpha-1,4 glucosidase activity in fibroblasts
GM4912	2	EEMEM 20U	0074	4MO	M	BL	--	A PROBAND			Biventric. hypertrophy, enlarged tongue, decreased muscle tone, slightly enlarged liver, resp. diff., fever, & cyanosis; expired at age 6 mo; neg. family hist.; def. acid-alpha-1,4 glucosidase activ.; normal debranching enzyme activity
Glycogen Storage Disease: Type III - Debranching Enzyme Deficiency - 23240											
GM0111	8	EEMEM 20U	0143	1	F	WH	--	A PROBAND			46,XX; less than 10% of normal debrancher enzyme activity
GM0226	12	EEMEM 20U	0143	13	F		--	A PROBAND			Less than 10% of normal debrancher enzyme activity

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Glycogen Storage Disease: Type III - Debranching Enzyme Deficiency - 23240												
GM0573	7	EEMEM 20U	0062	16	F	WH	--	B	PROBAND			Deficient debrancher enzyme activity
GM0576	6	EEMEM 20U	0062	7	F	OR	--	A	PROBAND			Chinese; deficient debrancher enzyme activity
GM0578	18	EEMEM 20U	0062	4	M	WH	--	B	PROBAND			Latin-American; deficient debrancher enzyme activity
GM2523	3	EEMEM 20U	0107	18MO	M	BL	--	A	PROBAND			Debrancher enzyme deficiency in muscle & low phosphorylase activity in liver; fibroblasts fail to form more glucose from a pnosphorylase limit dextrin than from glycogen
GM3388	7	EEMEM 20U	0077	18MO	M	WH	--	A	PROBAND			Arab; decreased amylo-1-6 glucosidase activity in fibroblasts
GM3390	6	EEMEM 20U	0077	16MO	M	WH	--	A	PROBAND			Arab; deficient amylo-1-6-glucosidase activity in fibroblasts
GM0303	9	EEMEM 20U	0095	9	F	WH	--	A	PROBAND	317	1	Formerly GM0055; less than 10% of normal debrancher enzyme activity
Glycogen Storage Disease: Type IV - Andersen Disease - 23250												
*GM0572	9	EEMEM 20U	0062	2	F	WH	--	A	PROBAND			Brancher enzyme deficiency in fibroblasts
Glycogen Storage Disease: Type V - McArdle Disease - 23260												
GM0577	7	EEMEM 20U	0130	39	M	WH	--		PROBAND			Absent muscle phosphorylase activity; normal activity in fibroblasts
Glycogen Storage Disease: Type VI - Hers Disease - 23270												
GM0579	9	EEMEM 20U	0062		M		--		PROBAND			Low hepatic phosphorylase activity; normal activity in fibroblasts
Glycogen Storage Disease: Type VIII, IX - P'ase Kinase Deficiency - 30600												
GM0575	5	EEMEM 20U	0062	3	M	WH	Y-	B	PROBAND			Mexican-American; less than 10% of normal phosphorylase b-kinase activity; type IX
GM1702	4	EEMEM 20U	0095	11	M		Y-		PROBAND			Normal debrancher enzyme activity in fibroblasts
Hyperpyruvicacidemia												
GM0583	3	EEMEM 20U	0038	7MO	F	WH			PROBAND			Hyperlacticacidemia
Lactic Acidosis - 24540												
*GM7711	8	EEMEM 20U	0006	60A	M	WH	--	B	PROBAND			Egyptian; parents are first cousins; EEG abnormalities; seizure activity; expired at age 1 month of respiratory failure; high lactate/pyruvate ratio; fibroblasts show decreased ability to oxidize pyruvate & abnormal NAD to NADH ratio

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Lactic Acidosis - 24540												
GM7819	7	EEMEM 20U	0519	2WK	F	WH	--		PROBAND			Severe metabolic acidosis; elevated serum lactate levels; biventricular cardiac hypertrophy; enlarged liver; expired at age 2 weeks
Leigh Encephalomyelopathy - 25600												
*GM1503A	5	EEMEM 20U	0045	42MO	F	WH	--		PROBAND			Infantile subacute; subnormal activation of pyruvate dehydrogenase complex in disrupted fibroblasts
GM3672	2	EEMEM 20U	0105	16MO	F	WH	--		PROBAND			Developmental regression; microcephalic; elevated blood lactic acid & pyruvate; only affected family member; 46,XX
Mannosidosis (Lysosomal Alpha-D-Mannosidase Deficiency) - 24850												
*GM0654	3	EEMEM 20U	0004	7	M	WH	--	A	PROBAND			Deficient alpha-mannosidase; Finnish
*GM2817	14	EEMEM 15U	0062	6	F	WH	--	A	PROBAND			22% of normal alpha-mannosidase activity
GM4518	11	EEMEM 20U	0379	2	F	WH	--	B	PROBAND			Severe growth failure; hypotonia; psychomotor retardation; mild hepatosplenomegaly; x-ray findings compatible with Hurler's syndrome; 8% of control fibroblast & 1% of control leukocyte acid-alpha-mannosidase activity
GM2051	18	EEMEM 20U	0077	7	F	WH	--	B	PROBAND	33	1	Variant; Arab; approximately 20% of normal alpha-mannosidase activity; similarly affected brother
GM2050	9	EEMEM 20U	0077	11	M	WH	--	B	AFF BRO	33	2	Variant; Arab; approximately 20% of normal alpha-mannosidase activity
GM2049	8	EEMEM 10U	0077	39	M	WH	+-(D)	B	FATHER	33	3	Arab; approximately 50% of normal alpha-mannosidase activity; two affected children
Mucolipidosis: Type I (Lipomucopolysaccharigosis) - 25240												
*GM2685	11	EEMEM 20U	0230	14	M	WH	--	A	PROBAND			Zero to 12% normal neuraminidase activity; normal B-glucuronidase and B-hexosaminidase levels; Hurler-like face, skeletal abnormalities, mental retardation, & cherry-red macular spot
Mucolipidosis: Type II - I-Cell Disease - 25250												
*GM3066	5	EEMEM 20U	0260	23FW	F	WH	--	A	PROBAND	34	1	Deficient fibroblast beta-galactosidase & N-acetylglucosaminylphosphotransferase activity; similarly affected brother

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF CARBOHYDRATE METABOLISM								
				Mucolipidosis: Type II - I-Cell Disease - 25250								
*GM0081	5	EEMEM 20U	0095	23	M	WH	+-(O)	A	FATHER	34	3	46,XY; two affected children; heterozygote level of N-acetylglucosaminylphosphotransferase
*GM0080	5	EEMEM 20U	0095	16	F	WH	+-(O)	A	MOTHER	34	4	46,XX; two affected children; heterozygote level of N-acetylglucosaminylphosphotransferase
*GM1586C	9	EEMEM 20U	0077	5WK	M	WH	--	A	PROBAND	35	1	Reduced B-galactosidase activity in skin fibroblasts; serum studies compatible with diagnosis; culture grows poorly
*GM1589	7	EEMEM 20U	0077		M	WH	+-(O)	A	FATHER	35	2	Heterozygote level of N-acetylglucosaminylphosphotransferase
*GM1590	7	EEMEM 20U	0077		F	WH	+-(O)	A	MOTHER	35	3	Heterozygote level of N-acetylglucosaminylphosphotransferase
*GM2013A	11	EEMEM 20U	0077	2WK	M	WH	--	A	PROBAND	36	1	Arab; fibroblasts show deficiencies for several lysosomal hydrolases; undetectable level of N-acetylglucosaminylphosphotransferase activity
GM2014	8	EEMEM 20U	0077	18	F	WH	+-(O)		MOTHER	36	2	Arab; clinically unaffected
*GM2046	6	EEMEM 20U	0095	25	F	WH	+-(O)	A	MOTHER	37	2	Two affected children; heterozygote level of N-acetylglucosaminylphosphotransferase
*GM2047	4	EEMEM 20U	0095	26	M	WH	+-(O)	A	FATHER	37	3	Two affected children; heterozygote level of N-acetylglucosaminylphosphotransferase
*GM3112E		R1640 20U	0260	21FW	F	WH	--	A	PROBAND	37	4	Brother (GM2045) was similarly affected; passage 4 at IMR; deficient fibroblast N-acetylglucosaminylphosphotransferase activity
*GM2273B	8	EEMEM 20U	0070	2	M	WH	--	A	PROBAND	38	1	Low levels of activity for several lysosomal enzymes; same patient as GM1423; undetectable level of N-acetylglucosaminylphosphotransferase activity
GM2274	3	EEMEM 20U	0070		F	WH	+-(O)		MOTHER	38	2	Clinically unaffected
				Mucolipidosis: Type III - Pseudo-Hurler Polydystrophy - 25260								
*GM0113	4	EEMEM 20U	0105	30MO	F	WH	--	A	PROBAND			Deficient fibroblast Alpha-L-iduronidase, B-glucuronidase, B-hexosaminidase, B-galactosidase, A-fucosidase, A-galactosidase, A-mannosidase, & GLcNAc phosphotransferase activity; complementation group A

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucolipidosis: Type III - Pseudo-Hurler Polydystrophy - 25260												
*GM1759A	3	EEMEM 20U	0092	13	M	WH	--	A	PROBAND			Similarly affected sister; deficient fibroblast B-hexosaminidase, B-galactosidase, B-glucuronidase, A-fucosidase, A-galactosidase, A-mannosidase, & GLcNAc phosphotransferase activity; complementation group A
*GM2065	3	EEMEM 20U	0126	9	M	WH	--	A	PROBAND			Deficient beta-galactosidase, hexosaminidase, arylsulfatase A, and beta-glucuronidase
*GM2425	6	EEMEM 20U	0134	15	M	WH	--	A	PROBAND			Deficient fibroblast B-hexosaminidase, B-galactosidase, A-fucosidase, A-galactosidase, A-mannosidase, neuraminidase, and GLcNAc phosphotransferase activity; excessive inclusion bodies in cytoplasm; complementation group A
*GM3685	4	EEMEM 20U	0126	15	M	WH	--	A	PROBAND			Abnormal mucopolysaccharide accumulation and deficiencies for multiple acid hydrolases in fibroblasts including B-galactosidase; deficient leuko & fibro N-acetylglucosaminylphosphotransferase & increased serum B-hexosaminidase
*GM2559	2	EEMEM 20U	0126	7	F	BL	--	A	PROBAND	501	1	Similarly affected sib; deficient fibroblast B-hexosaminidase, B-galactosidase, A-fucosidase, A-galactosidase, A-mannosidase, and N-acetylglucosaminylphosphotransferase activity; complementation group A
*GM2558	2	EEMEM 20U	0126	9	F	BL	--	B	AFF SIS	501	2	40% of normal beta-galactosidase & 15% of normal beta-hexosaminidase activity; deficient leukocyte N-acetylglucosaminylphosphotransferase and increased serum B-hexosaminidase
*GM3391	8	EEMEM 20U	0077	15	M	WH	--	A	PROBAND	610	1	Iranian; similarly affected sib; deficient fibroblast Alpha-L-iduronidase, arylsulfatase A, hexosaminidase A & B, B-galactosidase, A-fucosidase, A-galactosidase, and A-mannosidase activity; complementation group C; poss. variant
*GM3392	7	EEMEM 20U	0077	13	F	WH	--	B	AFF SIS	610	2	Iranian; deficient Alpha-L-Iduronidase, arylsulfatase A, alpha-mannosidase, hexosaminidase A & B, & beta-galactosidase activity; possible variant

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucolipidosis: Type III - Pseudo-Hurler Polydystrophy - 25260												
*GM1494	6	EEMEM 20U	0057	16	F	WH	--	A	PROBAND	1012	1	Deficient Alpha-L-Iduronidase, beta-glucuronidase, beta-galactosidase, & neuraminidase; 28% of normal level of N-acetylglucosaminylphosphotransferase activity
*GM7255	7	EEMEM 20U	0494	53	M	WH	+-(0)		FATHER	1012	2	Clinically normal
*GM7256	2	EEMEM 20U	0494	51	F	WH	+-(0)		MOTHER	1012	3	Clinically affected with hyperlipoproteinemia, type IV
Mucolipidosis: Type IV (Ganglioside Sialidase Deficiency) - 25265												
GM2525	22	EEMEM 20U	0077	2	F	WH	--	B	PROBAND			Ashkenazi; clinically affected
GM2526	15	EEMEM 20U	0077	2	F	WH	--	B	PROBAND			Ashkenazi; clinically affected
*GM2527	4	EEMEM 20U	0077	20MO	M	WH	--	A	PROBAND			Ashkenazi; about 25% of control neuraminidase activity employing GD1a & GD1b gangliosides
*GM2528	20	EEMEM 20U	0077	8MO	M	WH	--	B	PROBAND			Ashkenazi; fibroblasts show grossly abnormal storage bodies & normal levels of acid hydrolases
GM2629	9	EEMEM 20U	0077	2	M	WH	--		PROBAND			19% of control neuraminidase activity employing GD1b ganglioside
*GM2048	10	EEMEM 20U	0077	2	F	WH	--	B	PROBAND	229	1	Ashkenazi; see GM2533 Lymphoid; affected sib is GM2529 Amniotic; photophobia; bilateral corneal opacities; abnormal lysosomal storage bodies; about 28% of control neuraminidase activity employing GD1a and GD1b ganglioside
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
*GM0415	2	EEMEM 20U	0107	4	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
GM0887	5	EEMEM 20U	0057	7	F	WH	--	A	PROBAND			No detectable Alpha-L-Iduronidase activity
GM1053	5	EEMEM 20U	0057	4	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; culture grows slowly
GM1257	7	EEMEM 15U	0062	1	F	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; Latin-American
GM6214	8	EEMEM 20U	0107	3	M	WH	--	A	PROBAND			Corneal clouding; develop delay; enlarged head; gingival thickening; coarse features; excess MPS in urine; deficient Alpha-L-Iduronidase & excess MPS accumulation in fibro; def corrected to normal level by Hurler factor; 46,XY,t(5;11)pat

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
GM0003	7	EEMEM 20U	0095	26	F	WH	+-(0)		PARENT	39	2	46,XX; clinically normal mother of a child with Hurler syndrome; proband not in Repository
*GM0034	6	EEMEM 20U	0095	9MO	F	WH	--	A	PROBAND	40	1	Deficient Alpha-L-Iduronidase; 46,XX
GM0031	4	EEMEM 20U	0095	23	F	WH	+-(0)		MOTHER	40	2	46,XX; clinically unaffected
GM0042	4	EEMEM 20U	0095	25	M	WH	+-(0)		FATHER	40	3	46,XY; clinically unaffected
GM0798		EEMEM 20U	0062	1	F	WH	--	A	PROBAND	41	1	Deficient Alpha-L-Iduronidase; passage 3 at IMR
GM0799		EEMEM 20U	0062	22	F	WH	+-(0)		MOTHER	41	2	Passage 3 at IMR; clinically unaffected
GM0800		EEMEM 20U	0062	27	M	WH	+-(0)		FATHER	41	3	Passage 3 at IMR; clinically unaffected
*GM1391	2	EEMEM 20U	0070	9MO	F	WH	--	A	PROBAND	42	1	Deficient Alpha-L-Iduronidase
GM1392	3	EEMEM 20U	0070	24	F	WH	+-(0)		MOTHER	42	2	Clinically unaffected
GM1393	2	EEMEM 20U	0070	24	M	WH	+-(0)		FATHER	42	3	Clinically unaffected
Mucopolysaccharidosis: Type IH/S - Hurler/Scheie - 25280												
GM0512	4	EEMEM 20U	0008	15	F	IN	--	A	PROBAND			Deficient Alpha-L-Iduronidase
*GM0963	3	EEMEM 20U	0092	5	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
GM2846		EEMEM 20U	0096	14	F	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase; similarly affected sister; passage 5 at IMR
GM1254	9	EEMEM 20U	0062	4	M	WH	--	A	PROBAND	43	1	Deficient Alpha-L-Iduronidase; similarly affected sib
GM1255	6	EEMEM 15U	0062	10	M	WH	--	A	AFF BRO	43	2	Deficient Alpha-L-Iduronidase
GM1898	10	EEMEM 20U	0077	6	M	WH	--	A	PROBAND	44	1	Arab; deficient Alpha-L-Iduronidase
GM2016	6	EEMEM 10U	0077	35	F	WH	+-(0)		MOTHER	44	2	Arab; clinically unaffected
GM2017	6	EEMEM 20U	0077	40	M	WH	+-(0)		FATHER	44	3	Arab; clinically unaffected
GM2845	5	EEMEM 20U	0096	23	M	WH	--	A	PROBAND	326	1	Deficient Alpha-L-Iduronidase; similarly affected sib
GM2847	3	EEMEM 20U	0096	25	F	WH	--	A	AFF SIS	326	2	Deficient Alpha-L-Iduronidase
Mucopolysaccharidosis: Type IS - Scheie Syndrome - 25310												
GM1256	9	EEMEM 15U	0062	12	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type IS - Scheie Syndrome - 25310												
GM1323	3	EEMEM 20U	0092	58	M	WH	--	A	PROBAND			Deficient Alpha-L-Iduronidase
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990												
GM0039	4	EEMEM 20U	0066	9	M	WH	Y-	A	PROBAND			Accumulates mucopolysaccharide, correctable by Hunter factor; deficient iduronate sulfatase activity; 46,XY
GM0047	6	EEMEM 20U	0107	9	M	WH	Y-	A	PROBAND			Accumulates mucopolysaccharide, correctable by Hunter factor
GM0140	4	EEMEM 20U	0105	5	M	BL	Y-	A	PROBAND			Excessive mucopolysaccharide accumulation correctable by Hunter factor
GM0298	4	EEMEM 20U	0066	2	M	WH	Y-	A	PROBAND			Similarly affected brother; excessive mucopolysaccharide accumulation correctable by Hunter factor
*GM0690	3	EEMEM 20U	0092	14	M	WH	Y-	A	PROBAND			Deficient iduronate sulfatase activity
GM0862	2	EEMEM 20U	0107	11	M	WH	Y-	A	PROBAND			Mild type; similarly affected half brother; Puerto Rican; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1258	7	EEMEM 15U	0062	2	M	WH	Y-	A	PROBAND			Excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1929	5	EEMEM 20U	0096	7	M	WH	Y-	B	PROBAND			Severe type; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM2268A	9	EEMEM 20U	0168	5	F	WH	-?	A	PROBAND			Rare Hunter's in female; excessive mucopolysaccharide accumulation correctable by Hunter factor; 46,XX; deficient iduronate sulfatase activity
GM0614	3	EEMEM 20U	0092	18	M	WH	Y-	A	PROBAND	45	1	Mild type; deficient iduronate sulfatase activity
*GM0613	3	EEMEM 20U	0092		F	WH	+-(D)	A	MOTHER	45	2	Normal mucopolysaccharide accumulation; positive family history for MPS II
*GM0615	3	EEMEM 20U	0092	9	M	WH	Y-	A	PROBAND	46	1	Severe type; deficient iduronate sulfatase activity
*GM0620	3	EEMEM 20U	0092		F	WH	+-(D)	A	MOTHER	46	2	Normal mucopolysaccharide accumulation; two affected male children

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990												
GM1928	5	EEMEM 20U	0096	7	M	WH	Y-	B	PROBAND	48	1	Mild type; excessive mucopolysaccharide accumulation correctable by Hunter factor; similarly affected uncle
GM1927	5	EEMEM 20U	0096	16	M	WH	Y-	B	AFF UNCLE	48	2	Mild type; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1583	7	EEMEM 20U	0077	3	M	WH	Y-	A	PROBAND	247	1	Israeli; affected brother is GM1584 Amniotic; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM1896	9	EEMEM 20U	0088	20FW	M	WH	Y-	B	PROBAND	597	1	Same patient as GM1794; excessive mucopolysaccharide accumulation correctable by Hunter factor
GM3181A	3	EEMEM 20U	0170	22	M	WH	Y-	A	PROBAND	601	1	Excessive mucopolysaccharide accumulation correctable by Hunter factor; two similarly affected cousins
GM3180A	3	EEMEM 20U	0170	27	F	WH	+?	A	SIS	601	2	Normal mucopolysaccharide accumulation; possible heterozygote
Mucopolysaccharidosis: Type IIIA - Sanfilippo Syndrome, A - 25290												
GM0312	2	EEMEM 20U	0107	3	F	WH	--	A	PROBAND			Deficient heparan sulfate sulfatase
GM0629	3	EEMEM 20U	0066	10	M		--	A	PROBAND			Deficient heparan sulfate sulfatase, normal alpha-D-Glucosaminidase
GM0643	3	EEMEM 20U	0107	3	M	WH	--	A	PROBAND			Deficient heparan sulfate sulfatase
GM0934	2	EEMEM 20U	0070	7	F	WH	--	A	PROBAND			Deficient heparan sulfate sulfatase
*GM1881	7	EEMEM 20U	0140	3	M	WH	--	A	PROBAND			Deficient heparan N-sulfatase; normal Alpha-N-acetylglucosaminidase activity; 46,XY,t(1;21)(1qter>1p31::21q22>21qter; 21pter>21q22::1p31>1pter), balanced; excessive heparan sulfate in urine
GM6110	5	EEMEM 20U	0224	6	F	WH	--	B	PROBAND			Mental retardation; no language; spastic gait, coarse facies; typical roentgen bone findings; deficient heparan-N-sulfatase
GM0879	7	EEMEM 20U	0057	3	F	WH	--	A	PROBAND	49	1	Deficient heparan sulfate sulfatase
GM0886	7	EEMEM 20U	0057	19	F	WH	+-(O)		MOTHER	49	2	Clinically unaffected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type IIIA - Sanfilippo Syndrome; A - 25290												
GM1094	3	EEMEM 20U	0095	5	F	WH	--	A	PROBAND	50	1	Deficient heparan sulfate sulfatase; normal N-acetyl-alpha-glucosaminidase; same patient as GM1739
GM1739	3	EEMEM 20U	0095	6	F	WH	--	A	PROBAND	50	1	Same patient as GM1094; deficient heparan sulfate sulfatase; normal N-acetyl-alpha-glucosaminidase
GM1095	4	EEMEM 20U	0095		F	WH	+-(D)		MOTHER	50	2	Clinically unaffected
GM1096	3	EEMEM 20U	0095		M	WH	+-(D)		FATHER	50	3	Clinically unaffected
Mucopolysaccharidosis: Type IIIB - Sanfilippo Syndrome; B - 25292												
GM0156	5	EEMEM 20U	0107	7	M	WH	--	A	PROBAND			No detectable N-acetyl-alpha-D glucosaminidase activity; 46,XY
GM0737	3	EEMEM 20U	0107	7	M	WH	--	A	PROBAND			Deficient N-acetyl-alpha-glucosaminidase
*GM2552	3	EEMEM 20U	0105	7	F	WH	--	A	PROBAND			Deficient N-acetyl-alpha-glucosaminidase; increased urinary heparan sulfate
GM2931	5	EEMEM 20U	0077	3	F	WH	--		PROBAND			Ashkenazi; clinically affected
GM1426	3	EEMEM 20U	0070	1	F	WH	--	A	PROBAND	289	1	Deficient N-acetyl-alpha-D glucosaminidase; similarly affected brother
Mucopolysaccharidosis: Type IIIC - Sanfilippo Syndrome; C - 25293												
GM5157	6	EEMEM 20U	0088	8	M		--	B	PROBAND			Deficient Acetyl CoA:alpha-glucosaminide N-acetyltransferase activity
Mucopolysaccharidosis: Type IIID - Sanfilippo Syndrome; D - 25294												
*GM5093	7	EEMEM 20U	0402	7	M	IN	--		PROBAND			Mentally retarded & characteristic behavioral disturbances; excessive secretion of heparan sulfate & inability to release inorganic sulfate from N-acetyl-glucosamine-6-sulfate residues of heparan sulfate-derived oligosaccharides
Mucopolysaccharidosis: Type IV - Morquio Syndrome - 25300												
*GM0593	3	EEMEM 20U	0092	7	F	WH	--	A	PROBAND			Less than 1% of normal N-acetylgalactosamine 6-sulfate sulfatase; normal B-galactosidase & B-glucuronidase activity
GM0958	2	EEMEM 20U	0092	12	M	WH	--	A	PROBAND			Clinically atypical; 3% of normal N-acetylgalactosamine 6-sulfate sulfatase activity; normal B-galactosidase & B-glucuronidase activity

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DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type IV - Morquio Syndrome - 25300												
GM1259	9	EEMEM 15U	0062	14	F	WH	--	A	PROBAND			Clinically atypical; culture grows poorly; deficient N-acetylgalactosamine 6-sulfate sulfatase, beta-galactosidase, & beta-glucuronidase
GM1361	3	EEMEM 20U	0092	43	M	WH	--	A	PROBAND			Variant; 3% of normal N-acetylgalactosamine 6-sulfate sulfatase activity, normal B-galactosidase & B-glucuronidase
Mucopolysaccharidosis: Type IVB-Beta-Galactosidase Deficient Morquio - 25301												
GM3251	6	EEMEM 20U	0134	4	F	WH	--	A	PROBAND			Normal N-acetylgalactosamine 6-sulfate sulfatase activity; less than 4% of control B-galactosidase activity using either 4MU-B-gal or GM1 ganglioside
GM1602	3	EEMEM 20U	0066	11	F	WH	--	A	PROBAND	222	1	Normal level of N-acetylgalactosamine 6-sulfate sulfatase; less than 4% of control B-galactosidase activity using either 4MU-B-gal or GM1 ganglioside; similarly affected sister
GM2455	3	EEMEM 20U	0066	6	F	WH	--	A	AFF SIS	222	2	Normal level of N-acetylgalactosamine 6-sulfate sulfatase; normal beta-galactosidase (low normal)
GM2456	3	EEMEM 20U	0066	30	M	WH	+-(0)		FATHER	222	3	Two affected children
GM2555	4	EEMEM 20U	0066	30	F	WH	+-(0)		MOTHER	222	4	Two affected children
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320												
GM2849	3	EEMEM 20U	0070	35	M	WH	--	B	PROBAND			Deficient Arylsulfatase B
*GM0519	9	EEMEM 20U	0008	4	F	WH	--	A	PROBAND	51	1	See GM1022 Lymphoid; deficient Arylsulfatase B; short, coarse facies, prominent forehead, corneal opacities, lumbar kyphosis, stiff joints, umbilical hernia, mild hepatosplenomegaly, and dysostosis multiplex
*GM0520	8	EEMEM 20U	0008	42	M	WH	+-(0)	A	FATHER	51	2	Heterozygote level of Arylsulfatase B
*GM0935	6	EEMEM 20U	0008	40	F	WH	+-(0)	A	MOTHER	51	3	Heterozygote level of Arylsulfatase B
*GM0943	7	EEMEM 20U	0008	8	F	WH	+-	A	SIS	51	4	Heterozygote level of Arylsulfatase B
*GM0942	7	EEMEM 20U	0008	5	F	WH	++	A	SIS	51	5	Normal level of Arylsulfatase B; 46,XX
GM0538	9	EEMEM 20U	0105	18MO	F	WH	--	B	PROBAND	267	1	Deficient Arylsulfatase B, normal Alpha-L-Iduronidase; similarly affected sib

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				DISORDERS OF CARBOHYDRATE METABOLISM							
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320											
GM2572	3	EEMEM 20U	0105	2	M	WH	-- B	AFF BRO	267	2	Deficient Arylsulfatase B
GM3720	6	EEMEM 20U	0016	25	F	BL	+-(O)	MOTHER	392	1	See GM3721 Lymphoid; clinically unaffected
GM3722	3	EEMEM 20U	0016	40MO	F	BL	--	PROBAND	392	2	5% of normal Arylsulfatase B activity; see GM3723 Lymphoid
GM3724	5	EEMEM 20U	0016	41	M	BL	+-(O)	FATHER	392	3	See GM3725 Lymphoid; clinically unaffected
GM3726	3	EEMEM 20U	0016	9	F	BL	+?	SIS	392	4	Possible heterozygote; see GM3727 Lymphoid
Mucopolysaccharidosis: Type VII - Beta-Glucuronidase Deficiency - 25322											
GM3303	5	EEMEM 20U	0126	14DA	M		-- B	PROBAND			Deficient beta-glucuronidase
GM4668	9	EEMEM 20U	0126	3MO	F	WH	-- B	PROBAND			Severe infantile form with liver & heart failure; about 5% of control beta-glucuronidase activity; all other lysosomal enzyme activities are normal
*GM0121	4	EEMEM 20U	0126	3	M	BL	-- A	PROBAND	52	1	2% of normal B-glucuronidase, normal A-galactosidase, acid B-galactosidase, and A-mannosidase; 46,XY
*GM1550	3	EEMEM 20U	0126	28	M	BL	+-(O) A	FATHER	52	2	About 20% of normal beta-glucuronidase activity; clinically unaffected
*GM2074	4	EEMEM 20U	0126	21	F	BL	+-(O) A	MOTHER	52	3	About 20% of normal beta-glucuronidase activity; clinically unaffected
*GM0151	3	EEMEM 20U	0066	4	M	WH	-- A	PROBAND	425	1	Less than 5% of normal beta-glucuronidase activity; similarly affected sib
GM2784	4	EEMEM 20U	0066	3MO	F	WH	-- A	AFF SIS	425	2	No detectable beta-glucuronidase activity in fibroblasts
Neuraminidase Deficiency (Sialidoses Types I & II) - 25655											
*GM2837	6	EEMEM 20U	0134	31	M	WH	-- A	PROBAND			22% of normal sialidase activity; cherry-red spots; myoclonus without dementia; similarly affected brother died at age 33
*GM1718	2	EEMEM 20U	0070	2MO	F	WH	-- A	PROBAND	53	1	Less than 1% of normal neuraminidase activity; coarse facies; hepatosplenomegaly; dysostosis multiplex; normal mental development

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REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Neuraminidase Deficiency (Sialidoses Types I & II) - 25655												
*GM1719	3	EEMEM 20U	0070	25	F	WH	+-(0)	A	MOTHER	53	2	53% of normal neuraminidase activity employing 4-MU-NeuAc; 107% of control employing GD1a ganglioside
*GM1720	2	EEMEM 20U	0070	26	M	WH	+-(0)	B	FATHER	53	3	Low normal neuraminidase activity with fetuin as substrate; normal activity with MPN as substrate
GM2921	9	EEMEM 20U	0134	2	M	WH	--	A	PROBAND	330	1	6 times the normal level of sialic acid in fibroblasts; approx. 8% of normal sialidase activity
GM2922	11	EEMEM 20U	0134		M	WH	+-(0)	B	FATHER	330	2	58% of normal sialidase activity using NAN-lactose as substrate
GM2923	9	EEMEM 20U	0134	29	F	WH	+-(0)	B	MOTHER	330	3	62% of normal sialidase activity using NAN-lactose as substrate
Neuraminidase and Beta-Galactosidase Deficiency - 25654												
GM2438	12	EEMEM 20U	0049	9MO	F		--	A	PROBAND			From Canadian Repository; 20-30% of control B-galactosidase activity and 5% of control sialidase activity
*GM0806	3	EEMEM 20U	0126	30MO	F	WH	--	A	PROBAND	693	1	Deficient beta-galactosidase & sialidase activity; infantile onset; visceromegaly; dysostosis multiplex; normal stature & intelligence; see GM4305 Lymphoid
*GM4307	11	EEMEM 20U	0126	36	F	WH	+-(0)	A	MOTHER	693	2	Normal beta-galactosidase and neuraminidase activity; see GM6091 Lymphoid
*GM4306	6	EEMEM 20U	0126	36	M	WH	+-(0)	A	FATHER	693	3	Normal beta-galactosidase, neuraminidase & GM3 sialidase activity; normal sialic acid content; see GM6092A Lymphoid
*GM5076	23	EEMEM 20U	0401	7	M	WH	--	B	PROBAND	813	1	Coarse facies, hepatosplenomegaly, lumbar kyphosis, & no signs of neurological or ocular abnormalities; 10% of normal B-galactosidase & 6% of normal sialidase activity in fibroblasts
*GM5077	24	EEMEM 20U	0401	29	F	WH	+-(0)	B	MOTHER	813	2	Clinically normal; low normal fibroblast B-galactosidase activity; heterozygous level of sialidase activity; one affected child
Pyruvate Carboxylase Deficiency - 26615												
GM0444	3	EEMEM 20U	0019	20MO	M	WH	--	A	PROBAND			Lactic acidosis; about 2% of normal pyruvate carboxylase activity; 46,XY in leukocytes

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DISORDERS OF CARBOHYDRATE METABOLISM												
Pyruvate Carboxylase Deficiency - 26615												
*GM6056	5	EEMEM 20U	0436	4MO	F	WH	--	A	PROBAND			No detectable pyruvate carboxylase activity in fibroblasts; normal propionyl CoA carboxylase activity; severe lactic acidosis and failure to thrive
Phosphoglycerate Kinase Deficiency - 31180												
*GM0743	2	EEMEM 20U	0098	4	M	OR	Y-		PROBAND			Japanese; nonspherocytic hemolytic anemia; cerebral palsy; mental deficiency; decreased muscle & erythrocyte PGK activity; variant PGK (Matsue)
Salla Disease - 26874												
GM5520	2	EEMEM 20U	0319		F	WH	--	A	PROBAND	854	1	Yugoslavian; gargoyle-like features; hepatosplenomegaly; severe progressive mental & neurologic involvement; similarly affected brother; markedly elevated free sialic acid in urine & cultured skin fibro.; normal acid hydrolases
GM5521	2	EEMEM 20U	0319	24	F	WH	+-(O)	A	MOTHER	854	2	Yugoslavian; clinically normal; two affected children; normal sialic acid level in fibroblasts; 46,XX
GM5522	2	EEMEM 20U	0319	39	M	WH	+-(O)	A	FATHER	854	3	Yugoslavian; clinically normal; two affected children; normal sialic acid level in fibroblasts
Tricarboxylic Acid Cycle, Defect of - 27537												
*GM3093	9	EEMEM 20U	0088	4	F	WH	--	A	PROBAND			Pyruvate dehydrogenase complex deficiency (20% of control)
DISORDERS OF LIPID METABOLISM												
Apolipoprotein C-II Deficiency, Type 1 Hyperlipoproteinemia - 20775												
*GM3998	4	EEMEM 20U	0359	63	M	WH	--		PROBAND			Hypertriglyceridemia; chylomicronemia; pancreatitis; diabetes mellitus; parents are second cousins; positive family history for Apo C-II deficiency
Beta-sitosterolemia (Phytosterolemia; Hyperapobetalipoproteinemia) - 21025												
*GM1995	6	EEMEM 20U	0072	9	M	WH	--		PROBAND			Xanthomas
*GM1994	12	EEMEM 20U	0072	11	M	WH	--		PROBAND	352	1	Xanthomas; moderate hypercholesterolemia; similarly affected sib
GM1600	5	EEMEM 20U	0072	18	F	WH	--		AFF SIS	352	2	Tendon xanthomas; slightly elevated cholesterol

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						DISORDERS OF LIPID METABOLISM						
Beta-sitosterolemia (Phytosterolemia; Hyperapobetalipoproteinemia) - 21025												
GM1599	6	EEMEM 20U	0072	42	M	WH	+-(O)		FATHER	352	3	Diabetic; two affected children
GM1601	5	EEMEM 20U	0072	41	F	WH	+-(O)		MOTHER	352	4	Clinically normal; two affected children
Carnitine Deficiency, Systemic - 21214												
GM4081	7	EEMEM 20U	03o2	19MO	M	WH	--		PROBAND			Marked deficiency of carnitine in muscle, mild deficiency in liver; similarly affected fraternal twin; hypoglycemia; hepatic dysfunction; hyperbilirubinemia; ascites; numerous episodes of sepsis
Myopathy: Carnitine-palmityl-transferase Deficiency - 25511 & 25512												
GM1763		EEMEM 20U	0095	29	M	WH	--		PROBAND	344	1	Formerly GM0249; recurrent myoglobinuria; passage 4 at IMR
Cholesterol Ester Storage Disease of Liver - 21500												
GM3111	6	R1640 20U	0187	22	M	WH	--		PROBAND			Liver lysosomal acid lipase <5% of control and electrophoresis showed absence of "A" band
GM3347	8	EEMEM 20U	0006	7	F	WH	--	B	PROBAND			Mexican; approximately 5% of control level of acid lipase activity
*GM0863	5	EEMEM 20U	0006	11	F	WH	--	A	PROBAND	381	1	Latin American; two similarly affected sisters; less than 5% of normal acid lipase activity in fibroblasts; CRM+ with antibodies to acid lipase
Fabry Disease (Diffuse Angiokeratoma, Hereditary Dystopic Lipidosis) - 30150												
*GM0107	5	EEMEM 20U	0105	10	M		Y-	A	PROBAND			27% of normal level of alpha-galactosidase; normal beta-glucuronidase, acid-beta-galactosidase, and alpha-mannosidase
*GM0881	9	EEMEM 20U	0108	17	M	WH	Y-	B	PROBAND			About 20% of normal alpha-galactosidase activity; conjunctiva showed lipid deposits in the endothelial cells of the vessel walls
*GM0882	10	EEMEM 20U	0108	40	M	WH	Y-	A	PROBAND			Italian; history of proteinuria and mild fatigability; 13 to 30% of normal alpha-galactosidase activity
*GM2769	7	EEMEM 20U	0238	18	M	WH	Y-	A	PROBAND	246	1	Alpha gal A absent, Alpha gal B normal, Xg(a) antigen negative; positive family history; affected maternal grandfather
*GM2774	3	EEMEM 20U	0238	47	F	WH	+-(O)	A	MAT AUNT	246	3	50% of normal Alpha gal A, normal Alpha gal B; Xg(a) antigen +/-; two affected sons; two populations of cells, one with normal alpha-galactosidase A activity & a second with deficient enzyme activity

observed using cell sorting

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Fabry Disease (Diffuse Angiokeratoma, Hereditary Dystopic Lipidosis) - 30150												
*GM2775	3	EEMEM 20U	0238	16	M	WH	Y-	A	AFF MAT COU	246	4	Alpha gal A absent, Alpha gal B normal, Xg(a) antigen positive; 1st cousin to proband
*GM2771	6	EEMEM 20U	0238	42	F	WH	+-	A	MAT AUNT	246	5	50% of normal Alpha gal A, normal Alpha gal B, Xg(a) antigen +/-; two unaffected sons
*GM2772	4	EEMEM 20U	0238	38	F	WH	+-(O)	A	MAT AUNT	246	7	50% of normal Alpha gal A, normal Alpha gal B; Xg(a) antigen +/-; two affected sons; two populations of cells, one with normal Alpha-galactosidase A activity & a second with deficient enzyme activity observed using cell sorting
Farber Lipogranulomatosis; Ceramidase Deficiency - 22800												
GM2315	8	EEMEM 20U	0175	7	F	WH	--	B	PROBAND			Italian descent; a sister with similar clinical history died at age 18; deficient acid ceramidase
GM2314	4	EEMEM 20U	0175	6	F	WH	--	B	PROBAND	56	1	Irish descent; deficient acid ceramidase
GM2316	3	EEMEM 20U	0175	37	M	WH	+-(O)		FATHER	56	2	Irish descent; clinically unaffected
GM2317	3	EEMEM 20U	0175	37	F	WH	+-(O)		MOTHER	56	3	Irish descent; clinically unaffected
*GM5752	6	EEMEM 20U	0175	1DA	F	WH	--		PROBAND	862	1	Hepatosplenomegaly; nodules on hands and feet; pulmonary infiltrates; anemia; hoarse cry; adenopathy; elev. liver ceramide level; undetect. acid ceramidase activity in leukocytes & fibroblasts; similarly affected bro.; see GM5748 Lym.
GM1 Gangliosidosis: Type I; Beta - Galactosidase-1 Deficiency - 23050												
*GM0918A	20	EEMEM 20U	0049	1	M		--	A	PROBAND			Less than 1% of normal beta-galactosidase activity; 46,XY; from Canadian Repository; poor growing culture
GM5335	4	EEMEM 20U	0107	19MO	F	WH	--	B	PROBAND			Progressive dementia; progressive hypotonia; bony abnormality; retinal pigmentation; 1-2% of normal B-galactosidase activity in leukocytes & fibroblasts; normal sialidase activity
*GM3589	8	EEMEM 20U	0126	7MO	F	WH	--	A	PROBAND	808	1	Less than 1% of normal beta-galactosidase & normal sialidase activity; short stature; developmental delay; cherry-red spots; both parents have less than normal beta-galactosidase activity

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DISORDERS OF LIPID METABOLISM												
GM1 Gangliosidosis: Type I; Beta - Galactosidase-1 Deficiency - 23050												
GM5652	2	EEMEM 20U	0423	18MO	F	WH	--	A	PROBAND	856	1	Venezuelan; consang. parents; dev. delay; onset at 2-3 mos; hepatosplenomegaly; cherry-red spot; blind; resp. diff; norm. N-acetylglucosaminylphosphotransferase, B-hexosaminidase, B-glucuronidase & neur.; deficient B-galactosidase
GM5653	2	EEMEM 20U	0423	16MO	M	WH	--	A	AFF 1ST COU	856	2	Venezuelan; devel. delay; onset at 2-3 mos; hepatosplenomegaly; cherry-red spot; blindness; resp. diff.; norm. N-acetylglucosaminylphosphotransferase, B-hexosaminidase, B-glucuronidase, & neuraminidase; deficient B-galactosidase act
GM1 Gangliosidosis: Type III - 23065												
GM2439	16	EEMEM 20U	0049		M		--	A	PROBAND			From Canadian Repository; deficient B-galactosidase activity; 46,XY
GM2 Gangliosidosis: Tay-Sachs Disease; AB Variant - 27275												
*GM1675	5	EEMEM 20U	0008	16MO	F	BL	--		PROBAND			Normal B-hexosaminidase A alpha and beta chains
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
*GM0077	9	EEMEM 20U	0095	1	M	BL	--	A	PROBAND			Non-Jewish; deficient hexosaminidase A
*GM0221	4	EEMEM 20U	0066	3	M	WH	--	A	PROBAND			Deficient hexosaminidase A
*GM0502B	5	EEMEM 20U	0066	11MO	M	WH	--	A	PROBAND			Jewish; deficient hexosaminidase A; electrophoresis shows absence of A band
GM0527	3	EEMEM 20U	0107	15MO	M	WH	--	A	PROBAND			Deficient hexosaminidase A
*GM4863	2	EEMEM 20U	0066	22MO	M	WH	--?		PROBAND			Ashkenazi, clinically normal; possible variant; 29% normal hexosaminidase A in leukocytes, 9% in fibroblasts, & none in serum or plasma; both parents have heterozygous levels of hexosaminidase A
*GM0515A	8	EEMEM 20U	0068	1	F	WH	--	A	PROBAND	58	1	Pericardium fibroblast culture; Jewish; similarly affected sib; deficient hexosaminidase A activity; electrophoresis shows absence of A band; no detectable alpha chain synthesis; no detectable alpha chain mRNA
GM0514	8	EEMEM 20U	0068	3	F	WH	--	B	AFF SIS	58	2	Pericardium fibroblast culture; Jewish; deficient hexosaminidase A

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DISORDERS OF LIPID METABOLISM												
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
*GM1110	7	EEMEM 20U	0068	10MO	M	WH	--	A	PROBAND	59	1	Non-Jewish; electrophoresis shows absence of hexosaminidase A band; consanguineous parents; synthesizes an altered alpha chain polypeptide
GM1109A	9	EEMEM 20U	0068	31	M	WH	+-(O)		FATHER	59	2	Non-Jewish; clinically unaffected
GM1108A	9	EEMEM 20U	0068	30	F	WH	+-(O)	A	MOTHER	59	3	Non-Jewish; clinically unaffected
*GM2968	5	EEMEM 20U	0008	12MO	F	WH	--	A	PROBAND	437	1	Ashkenazi; no detectable hexosaminidase A activity by electrophoresis; no detectable alpha chain synthesis; no detectable alpha chain mRNA
GM3051	8	EEMEM 20U	0008	23	M	WH	+-(O)	B	FATHER	437	2	Ashkenazi; clinically unaffected
GM3052	7	EEMEM 20U	0008	24	F	WH	+-(O)	B	MOTHER	437	3	Ashkenazi; same patient as GM2969; clinically unaffected
GM2 Gangliosidosis: Type II - Sanghoff Disease - 26880												
*GM0294	5	R1640 20U	0017	2	M	WH	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0317A	7	EEMEM 20U	0066	1	F	BL	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0470	8	EEMEM 20U	0009	1	F	WH	--	A	PROBAND			Deficient hexosaminidase A & B
*GM0203A	8	EEMEM 20U	0107	1	M	WH	--	A	PROBAND	60	1	Deficient hexosaminidase A & B; 46,XY
GM0204	5	EEMEM 20U	0107	18	F	WH	+-(O)		MOTHER	60	2	46,XX; clinically unaffected
*GM2094A	4	EEMEM 20U	0181	12	M	WH	--	B	PROBAND	432	1	Juvenile; cerebellar ataxia; severe mental retardation; deficient hexosaminidase A and B; same patient as GM2144
Gaucher Disease: Type I (Non-cerebral, Acid Beta-Glucosidase Def.) - 23080												
*GM1607	3	EEMEM 20U	0047	30	M	WH	--	A	PROBAND			Adult; eastern European Jewish; hepatosplenomegaly since age 17; typical Gaucher's cells observed in bone marrow aspirant; negative family history; 18% of control fibroblast glucocerebrosidase activity
*GM4394	2	EEMEM 20U	0126	16MO	M	BL	--	A	PROBAND			Hepatosplenomegaly; bone pain; no family history of disease; no neurologic abnormalities at age two; 5% of control fibroblast glucocerebrosidase activity
GM7968	4	EEMEM 20U	0526	19FW	M	WH	--		PROBAND			Fetal skin fibroblast culture; positive family history; deficient Beta-glucosidase activity in cultured amniotic fluid cells

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DISORDERS OF LIPID METABOLISM												
Gaucher Disease: Type II (Infantile, Cerebral) - 23090												
*GM1260	2	EEMEM 20U	0050	11MO	F	WH	--	A	PROBAND			Splenomegaly; opisthotonos; expired at age 1; 3% of control fibroblast glucocerebrosidase activity
*GM2627	2	EEMEM 20U	0107	3	F	WH	--	A	PROBAND			Atypical; subacute neuropathic Gauchers; myoclonus; seizures; expired at age 3 due to pneumonia; 24% of control fibroblast glucocerebrosidase activity
*GM0877	2	EEMEM 20U	0107	1	M	WH	--	A	PROBAND	57	1	Hepatosplenomegaly; strabismus; trismus; expired at age 1; 4% of control fibroblast glucocerebrosidase activity
GM0878	2	EEMEM 20U	0107		F	WH	+-(O)	A	MOTHER	57	2	Carrier level of fibroblast glucosylceramide B-glucosidase activity
Gaucher Disease: Type III (Juvenile and Adult, Cerebral) - 23100												
*GM0372	4	EEMEM 20U	0009	29	M	WH	--	A	PROBAND			Adult; 6% of control fibroblast glucocerebrosidase activity
*GM0852	5	EEMEM 20U	0138	20	M	WH	--	A	PROBAND			Adult; 6% of control fibroblast glucocerebrosidase activity
Hepatic Lipase Deficiency - Hyperlipoproteinemia												
*GM6946	3	EEMEM 20U	0359	48	M	WH			PROBAND	976	1	Increased LDL and IDL; skin xanthomata; coronary heart disease with diabetes mellitus; positive family history; uncertain inheritance; see GM6945 Lymphoid
Familial Hyperlipoproteinemia: Type I (Lipoprotein Lipase Def.) - 23860												
GM6978	2	EEMEM 20U	0006	10MO	M	WH	--		PROBAND			Cutaneous xanthomas; hypertriglyceridemia; similarly affected brother; lipoprotein lipase deficiency; no apolipoprotein C-II deficiency; normal cholesterol level; conc. of low & high density lipoproteins is depressed
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
*GM1915	4	EEMEM 20U	0050	13	F	BL	--	A	PROBAND			Receptor negative
*GM0283	5	EEMEM 20U	0045	28	F	WH	+-		MOTHER	61	1	Formerly GM0102; elevated plasma cholesterol
*GM0376	5	EEMEM 20U	0045	6	F	WH	+-		PROBAND	61	2	Formerly GM0099; elevated plasma cholesterol
*GM0488A	9	EEMEM 20U	0084	11	M	WH	--	A	PROBAND	62	1	Receptor negative; approx. 10 fold increase in HMG CoA reductase in fibroblasts

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DISORDERS OF LIPID METABOLISM												
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
*GM0483	4	EEMEM 20U	0084	37	F	WH	+-		MOTHER	62	2	Elevated plasma cholesterol
*GM2000B	3	EEMEM 20U	0050	7	F	WH	--	A	PROBAND	63	1	Receptor negative; same patient as GM1116; LDL receptor activity < 1% of value obtained with normal cells
*GM0701A	10	EEMEM 20U	0072	6	F	WH	--	A	PROBAND	64	1	Receptor negative; approx. 10 fold increase in HMG CoA reductase in fibroblasts; LDL receptor activity < 1% of value obtained with normal cells
*GM0700A	11	EEMEM 20U	0072	45	F	WH	+-	A	MOTHER	64	2	LDL receptor activity approximately 36% of value obtained with control fibroblasts
*GM2408A	2	EEMEM 20U	0050	16	M		--	A	PROBAND	272	1	Normal receptor binding with defective receptor internalization; line J.D.; internalization index is 2 vs 17-48 for normocholesterolemic fibroblast cultures
GM1355	3	EEMEM 20U	0093	13	F	WH	-?	A	PROBAND	274	1	25 to 40% of normal LDL receptor activity; possibly a severe heterozygote, mild homozygote, or double heterozygote
GM1354	3	EEMEM 20U	0093	38	F	WH	+-		MOTHER	274	2	Elevated plasma cholesterol; no xanthomas
GM1385	3	EEMEM 20U	0093	38	M	WH	+-		FATHER	274	3	Elevated plasma cholesterol; no xanthomas
GM1386	2	EEMEM 20U	0093	17	M	WH	++		BRO	274	4	Normal plasma cholesterol; 46,XY; 14% of cells are tetraploid at passage 5
*GM0486	4	EEMEM 20U	0084	25	F	WH	--	A	PROBAND	387	1	Receptor negative; see GM1459A Lymphoid
*GM3040B	16	EEMEM 20U	0072	9	M	WH	--	A	PROBAND	440	1	Receptor negative or defective; very high serum LDL; normal triglycerides; cutaneous and tendinous xanthomas; corneal arcus
GM3064	6	EEMEM 20U	0072	29	M	WH	+-		FATHER	440	2	No xanthomas; no clinical evidence of atherosclerosis; elevated plasma cholesterol
GM3065	6	EEMEM 20U	0072	30	F	WH	+-		MOTHER	440	3	No xanthomas; no clinical evidence of atherosclerosis; family history is strongly positive for early atherosclerosis and hypercholesterolemia

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DISORDERS OF LIPID METABOLISM												
Familial Hypercholesterolemia - Variants												
*GM0667	5	EEMEM 20U	0072	12	M	WH	--		PROBAND	263	1	Lebanese; elevated plasma cholesterol; pedigree shows autosomal recessive inheritance; three affected sibs
*GM0694	6	EEMEM 20U	0072	15	F	WH	--		AFF SIS	263	2	Lebanese; elevated plasma cholesterol; xanthomas
GM0695	6	EEMEM 20U	0072	55	M	WH	+-(0)		FATHER	263	3	Lebanese; four affected children
*GM0696	6	EEMEM 20U	0072	5MO	F	WH	--		AFF SIS	263	4	Lebanese; elevated plasma cholesterol; xanthomas
*GM0697	6	EEMEM 20U	0072	17	M	WH	--		AFF BRO	263	5	Lebanese; elevated plasma cholesterol; xanthomas
Krabbe Disease (Globoid Cell Sclerosis) - 24520												
GM1773	2	EEMEM 20U	0105	1	M	WH	--	A	PROBAND			Deficient lactosylceramide beta-galactosidase
GM4517	2	EEMEM 20U	0292	8MO	M	WH	--		PROBAND			Developmental delay; seizures and spasticity; deficient galactocerebrosidase activity in WBC
GM0267	7	R1640 20U	0058	36	F	WH	+-(0)	A	PARENT	97	1	Proband not in Repository; 35% of normal galactocerebroside beta-galactosidase activity
GM0268	6	R1640 20U	0058	41	M	WH	+-(0)	A	PARENT	97	2	Proband not in Repository; 45% of normal galactocerebroside beta-galactosidase activity
GM3045	2	EEMEM 20U	0008	19FW	F		--		PROBAND	442	1	Identical twin (one placenta, one sac) of GM3046A; clinically affected
GM3046A		EEMEM 20U	0008	19FW	F		--		IDENT TWIN	442	2	Affected; identical twin (one placenta, one sac) of GM3045; passage 4 at IMR
GM4372	5	EEMEM 20U	0107	16FW	M		--		PROBAND	716	1	Pedigree includes a previously affected sib; deficient galactosyl and lactosyl ceramide beta-galactosidase activity in cultured amniotic fluid cells
GM4913	4	EEMEM 20U	0224	21FW		WH	--	B	PROBAND	778	1	Spleen biopsy; positive family history; almost no detectable galactosylceramide B-galactosidase activity in brain, spleen, & cultured amniotic fluid cells; same patient as GM4914

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DISORDERS OF LIPID METABOLISM												
Krabbe Disease (Globoid Cell Sclerosis) - 24520												
GM4914	3	EEMEM 20U	0224	21FW		WH	--	B	PROBAND	778	1	Brain biopsy; positive family history; almost no detectable galactosylceramide B-galactosidase activity in brain, spleen, & cultured amniotic fluid cells; same patient as GM4913
GM6806	2	EEMEM 20U	0458	2	F	WH	--		PROBAND	962	1	Seizures; cortically blind; developmental delay; poor feeding; onset at age 6 months; deficient WBC lactosylceramide B-galactosidase activity; see GM6805 Lymphoid
GM6810	2	EEMEM 20U	0458		M	WH	+-(O)		FATHER	962	2	Clinically normal; heterozygote level of lactosylceramide B-galactosidase activity in WBC; see GM6807A Lymphoid
GM6808	2	EEMEM 20U	0458		F	WH	+-(O)		MOTHER	962	3	Clinically normal; heterozygote level of lactosylceramide B-galactosidase activity in WBC; see GM6809 Lymphoid
Long-Chain Acyl CoA Dehydrogenase Deficiency												
GM6127	6	EEMEM 20U	0187	3MO	M	WH	--	B	PROBAND			Carnitine def. in plasma & liver; persistent cardiomyopathy; skel. muscle weakness; fasting hypoglycemia; def. of leukocyte long-chain Acyl CoA dehydrogenase; fibro. have profoundly reduced oxidation of long-chain fatty acids
GM7843	14	EEMEM 20U	0187	10MO	F	WH	--		PROBAND			Episodes of vomiting; diarrhea, lethargy, hypoglycemia, & dicarboxylicaciduria; low plasma carnitine levels; 10% of normal long-chain Acyl CoA dehydrogenase activity in leukocytes
Medium-Chain Acyl CoA Dehydrogenase Deficiency												
*GM4488	5	EEMEM 20U	0362		F	WH	--	A	PROBAND			Deficiency of serum & muscle carnitine; acute episodes of encephalopathy associated with hepatic dysfunction & progressive muscle weakness; deficient medium-chain Acyl CoA dehydrogenase activity
*GM5872	14	EEMEM 20U	0187	1	F	WH	--		PROBAND			Coma; dicarboxylicaciduria; low plasma carnitine level; fibroblasts show defect in fatty acid oxidation due to medium-chain Acyl CoA dehydrogenase deficiency; non-ketotic hypoglycemia
GM7842	19	EEMEM 20U	0187	5	F	WH	--	B	PROBAND			Reyes syndrome-like episodes with hypoglycemia; low plasma & tissue carnitine levels; dicarboxylicaciduria; failure of ketosis; mild hyperammonemia; 13% of normal medium-chain Acyl CoA dehydrogenase activity in fibroblasts

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DISORDERS OF LIPID METABOLISM												
Medium-Chain Acyl CoA Dehydrogenase Deficiency												
GM7844	18	EEMEM 20U	0187	9MO	M	WH	--	B	PROBAND			Recurrent episodes of hypoglycemia; non-ketotic coma with dicarboxylicaciduria; low plasma carnitine levels; 12% of normal medium-chain Acyl CoA dehydrogenase activity in fibroblasts
*GM5870	6	EEMEM 20U	0187	2	F	WH	--		PROBAND	882	1	Coma; seizures; fatty liver; low levels of carnitine in plasma & tissues; fibroblasts show defect in fatty acid oxidation due to medium-chain Acyl CoA dehydrogenase deficiency; similarly affected sib; non-ketotic hypoglycemia
GM6126	10	EEMEM 20U	0187	34	F	WH	+-(0)		MOTHER	882	3	Clinically unaffected; two affected children; 50% of normal medium-chain Acyl CoA dehydrogenase activity in leukocytes; slow growing culture
GM6125	6	EEMEM 20U	0187	16	F	WH	--		PROBAND	1058	1	Recurrent coma; dicarboxylicaciduria; less than 10% of normal medium-chain Acyl CoA dehydrogenase activity in leukocytes; non-ketotic hypoglycemia; see GM7523 Lymphoid
Metachromatic Leukodystrophy: Adult - 25000												
GM0132A	12	EEMEM 20U	0105	22	M	WH	+-	A	1ST COU	66	1	Heterozygote levels of arylsulfatase A
GM2093	3	EEMEM 20U	0105	25	M	WH	--	A	PROBAND	66	3	Deficient arylsulfatase A
Metachromatic Leukodystrophy: Infantile (Arylsulfatase A Deficiency) - 25010												
*GM0078	7	EEMEM 20U	0131	3	M	WH	--	A	PROBAND			Deficient arylsulfatase A
GM0243	2	EEMEM 20U	0074	3	M	WH	--	A	PROBAND			Deficient arylsulfatase A
*GM0905	2	EEMEM 20U	0122	3	F	WH	--	A	PROBAND			Deficient arylsulfatase A
GM2331	5	EEMEM 20U	0134	5	M	WH	--	A	PROBAND			Deficient arylsulfatase A
*GM0197	3	EEMEM 20U	0105	4	M	WH	--	A	PROBAND	65	1	Deficient arylsulfatase A; two similarly affected sibs
*GM0196	3	EEMEM 20U	0105	33	M	WH	+-(0)	A	FATHER	65	2	46,XY; three children affected with infantile MLD, heterozygous level of arylsulfatase A in fibroblasts
GM0200	4	EEMEM 20U	0105	5	M	WH	+-	B	BRO	65	4	46,XY; carrier level of arylsulfatase A
GM0357		EEMEM 20U	0105	3	M	WH	+-	B	MAT 1ST COU	65	6	Carrier level of arylsulfatase A; passage 4 at IMR

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				DISORDERS OF LIPID METABOLISM							
Metachromatic Leukodystrophy: Infantile (Arylsulfatase A Deficiency) - 25010											
GM0640	10	EEMEM 20U	0105	25	F	WH	+-	B MAT AUNT	65	8	Mother of GM0357; carrier level of arylsulfatase A
Multiple Sulfatase Deficiency Disease - 27220											
*GM4681	6	EEMEM 20U	0134	1	M	WH	--	A PROBAND			Severe mental retard.; dysostosis multiplex; ichthyotic rash; arylsulfatase A in fibroblasts 42% of normal, but less than 5% in urine, leukocytes, and serum; elevated urinary sulfatides and MPS
*GM2407A	10	EEMEM 20U	0214	3	F	WH	--	A PROBAND	237	1	Deficient arylsulfatase A, B, & C; sulfamidase, iduronate-sulfate sulfatase, GalNAc-6 and GlcNAc-6-sulfate sulfatase
GM2644	7	EEMEM 20U	0214	22	F	WH	+-(O)	MOTHER	237	2	Clinically unaffected
GM2667	7	EEMEM 20U	0214	27	M	WH	+-(O)	FATHER	237	3	Clinically unaffected
*GM3245	3	EEMEM 20U	0105	9	M	WH	--	A PROBAND	632	1	See GM3246 Lymphoid; microcephaly, delayed psychomotor develop., hypotonia, blind, deaf, & myoclonus; excess heparan sulfate in urine; deficient leukocyte arylsulfatase A,B,& C; reduced arylsulfatase A,B,& C activity in fibroblasts
Niemann-Pick Disease: Type A (Sphingomyelinase Deficiency) - 25720											
*GM0112	3	EEMEM 20U	0143	10MO	M	WH	--	A PROBAND			No detectable sphingomyelinase activity and increased cholesterol biosynthesis in fibroblasts
*GM0370	8	EEMEM 15U	0002	13MO	F	WH	--	A PROBAND			7% of normal sphingomyelinase activity, deficient phospholipase C activity, and increased cholesterol biosynthesis in fibroblasts
*GM0406	9	EEMEM 20U	0105	21MO	F	WH	--	A PROBAND			8% of normal sphingomyelinase activity & deficient phospholipase C activity in fibroblasts
*GM0559		EEMEM 20U	0105	25	F	WH	+-	B PROBAND			Ashkenazi; clinically unaffected; positive family history; about 20% of normal sphingomyelinase activity & increased cholesterol biosynthesis in fibroblasts; passage 4 at IMR
GM0641	19	EEMEM 20U	0105	4	M	WH	--	B PROBAND			Deficient sphingomyelinase activity
Niemann-Pick Disease: Type B (Sphingomyelinase Deficiency) - 25720											
*GM3252	3	EEMEM 20U	0095	21FW	F	WH	--	A PROBAND			2% of normal sphingomyelinase activity & deficient phospholipase C activity in fibroblasts; similarly affected brother

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DISORDERS OF LIPID METABOLISM												
Niemann-Pick Disease: Type B (Sphingomyelinase Deficiency) - 25720												
*GM3393	8	EEMEM 20U	0006	10	F	WH	--	A	PROBAND			Non-Jewish; less than 5% of normal sphingomyelinase activity & deficient phospholipase C activity in fibroblasts
Niemann-Pick Disease: Type C (Sphingomyelinase Deficiency) - 25720												
*GM0110A	6	EEMEM 20U	0107	9	M	WH	--	A	PROBAND			Variant; 24% - 50% of normal sphingomyelinase activity; a brother died of Niemann-Pick at age 11
*GM3123	2	EEMEM 20U	0107	9	F	WH	--	A	PROBAND	451	1	See GM3124 Lymphoid; 38% of normal sphingomyelinase activity and normal B-galactosidase activity in fibroblasts
Niemann-Pick Disease: Type Unspecified												
GM0165	3	EEMEM 20U	0107	7	M	WH	--	A	PROBAND			Clinically atypical; 27% of normal sphingomyelinase activity
GM2895	6	EEMEM 20U	0073	2	M	WH	--	A	PROBAND	328	1	47% of normal sphingomyelinase activity; juvenile or variant form; Puerto Rican; clinically affected
GM2896	5	EEMEM 20U	0073	33	F	WH	+-(O)	B	MOTHER	328	2	50% of normal sphingomyelinase activity; clinically normal
GM2897	8	EEMEM 20U	0073	25	M	WH	+-(O)	B	FATHER	328	3	50% of normal sphingomyelinase activity; clinically normal
Niemann-Pick Disease without Sphingomyelinase Deficiency - 25725												
GM3519	14	EEMEM 20U	0333	29	M	WH	--	A	PROBAND			Normal sphingomyelinase activity; not Jewish or Nova Scotian; two similarly affected male sibs; sea blue histiocytes in bone marrow; very slow growing culture
Refsum Syndrome; Phytanic Acid Oxidase Deficiency - 26650												
*GM3896	9	EEMEM 20U	0175	38	M	WH	--	B	PROBAND	574	1	Elevated phytanic acid in blood, fat, & other body fluids and cultured skin fibroblasts; retinitis pigmentosa; see GM3897 Lymphoid
Schilder Disease - 26910 + 30010												
GM0269	4	EEMEM 20U	0058	30	F	WH	+?		PARENT			Possible carrier for fatal X-linked Schilder's disease with adrenal insufficiency; one affected son
*GM2448	7	EEMEM 15U	0175	8	M	WH	--		PROBAND			Adrenoleukodystrophy; mild neurological impairment - not clearly progressive; negative family history; four normal maternal uncles

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Sea-Blue Histiocyte Disease - 26960												
*GM0843	3	EEMEM 20U	0066	10	F	WH	--		PROBAND	121	1	Similarly affected sib; normal fibroblast activity for phospholipase A1, A2, & C, acid lipase, and sphingomyelinase
GM0844	3	EEMEM 20U	0066		M	WH	--		AFF BRO	121	2	Similarly affected sister
GM1912	3	EEMEM 20U	0348	24	F	WH	--		PROBAND	407	1	See GM1913 Lymphoid; muscle biopsy showed foamy histiocytes & sea-blue histiocytes; eye movement abnormalities
Wolman Disease; Lysosomal Acid Lipase Deficiency - 27800												
*GM1606	4	EEMEM 20U	0154	7MO	F		--	A	PROBAND			Deficient lysosomal acid lipase activity; CRM+ with antibodies to acid lipase
*GM2109	11	EEMEM 20U	0178	3MO	M	BL	--	A	PROBAND	631	1	Deficient lysosomal acid lipase activity; CRM+ with antibodies to acid lipase; 46,XY,5q+ in original culture; 46,XY in Repository stock; slow growing culture
GM3557	6	EEMEM 20U	0178	24	F	BL	+?	B	MOTHER	631	2	Possible carrier levels of acid lipase; 46,XX
GM3558	5	EEMEM 20U	0178	28	M	BL	+?	B	FATHER	631	3	Normal level of acid lipase; 46,XY
GM6122	11	EEMEM 20U	0187	30	M	WH	+-(O)	B	FATHER	909	1	45% of normal lysosomal acid lipase activity
GM6123	9	EEMEM 20U	0187	25	F	WH	+-(O)	B	MOTHER	909	2	50% of normal lysosomal acid lipase activity
GM6144	8	EEMEM 20U	0187	2MO	M	WH	--	B	PROBAND	909	3	Hepatosplenomegaly; adrenal calcification; deficient lysosomal acid lipase activity
MUTATIONS OF THE HEMOGLOBIN LOCI												
Sickle Cell Anemia (Hemoglobin S) - 14190												
GM2340	10	EEMEM 10I	0088	20FW	F		--		PROBAND			Sickle cell disease
Sickle Cell Anemia - Hb Kenya Compound Heterozygote												
*GM6342	5	EEMEM 20U	0457	36	M	BL	+-		PROBAND			Kenyan; healthy; HbS-Hb Kenya-gamma(G)-HPFH condition; mother has sickle cell trait; non-alpha chain of Hb Kenya shows amino terminal segment seq.(1-80) of the gamma chain of HbF and carboxy terminal segment (87-146) of beta chain of HbA
Thalassemias - 27350 and Hereditary Persistence of Fetal Hemoglobin - 14247												
*GM1368	4	EEMEM 20U	0096	15	M	BL	--	B	PROBAND			Well, but has 100% fetal hemoglobin; shows substantial deletion of delta and beta globin genes on both chromosomes 11; 46,XY

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MUTATIONS OF THE HEMOGLOBIN LOCI												
Thalassemias - 27350 and Hereditary Persistence of Fetal Hemoglobin - 14247												
GM3433	7	EEMEM 20U	0049	29FW	F	OR	--	A	PROBAND			Chinese; Canadian Rep. No. 646; electrophoresis confirmed Barts Hb as major component; alpha-thalassemia; positive family history; doesn't hybridize alpha-globin cDNA probe; 46,XX
GM4930	2	EEMEM 20U	0183	17MO	M	BL	--		PROBAND			Gamma(A)+ thalassemia homozygote; nearly 100% gamma(G) in cord blood HbF; percentage was consistently greater than 90% up to 300 days after birth; 46,XY; 12% of cells show random chromosome loss
GM3037	5	EEMEM 20U	0088	23FW	M	OR	--	A	PROBAND	948	1	Alpha-thalassemia; fetal hydrops; Vietnamese; confirmed by cDNA hybridization and globin chain synthesis; skin fibroblast culture; same patient as GM3038; 46,XY
GM3038	8	EEMEM 20U	0088	23FW	M	OR	--	B	PROBAND	948	1	Alpha-thalassemia; fetal hydrops; Vietnamese; confirmed by cDNA hybridization and globin chain synthesis; lung fibroblast culture; same patient as GM3037
DISORDERS OF METAL METABOLISM												
Acrodermatitis Enteropathica - 20110												
GM2814	3	EEMEM 20U	0070	6MO	M	WH	--		PROBAND	323	1	Low serum zinc concentration; clinically affected
GM2815	3	EEMEM 20U	0070	24	M	WH	+-(O)		FATHER	323	2	Heterozygote serum zinc concentration; clinically unaffected
Menkes Syndrome (Kinky Hair Disease) - 30940												
*GM0220	4	EEMEM 20U	0107	2MO	M	WH	Y-	A	PROBAND	79	1	Fibroblasts exhibit elevated Cu levels; positive family history; similarly affected brother and cousin; abnormal metallothionein gene regulation in response to copper
*GM1057	3	EEMEM 20U	0107	5DA	M	WH	Y-	A	AFF BRO	79	2	See GM1245 Lymphoid; fibroblasts exhibit elevated copper concentrations; 46,XY; abnormal metallothionein gene regulation in response to copper
*GM0245	3	EEMEM 20U	0074	1MO	M	WH	Y-	A	AFF COU	79	3	Fibroblasts exhibit elevated Cu levels; 46,XY; abnormal metallothionein gene regulation in response to copper

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF METAL METABOLISM												
Menkes Syndrome (Kinky Hair Disease) - 30940												
*GM4068	3	EEMEM 20U	0107		F	WH	+-(D)		AUNT	79	4	Mother of GM0245; 1 affected son; 2 affected nephews; abnormal (heterozygote) metallothionein gene regulation in response to copper
*GM1981	3	EEMEM 20U	0107	2	M	WH	Y-	A	PROBAND	80	1	See GM1982 Lymphoid; fibroblasts exhibit elevated copper concentrations; abnormal metallothionein gene regulation in response to copper
*GM1983	3	EEMEM 20U	0107	26	F	WH	+?		MOTHER	80	2	See GM1984 Lymphoid; son is only affected child in an extended family; normal metallothionein gene regulation in response to copper
GM3700	2	EEMEM 20U	0078	9MO	M	WH	Y-	A	PROBAND	390	1	Low serum copper and ceruloplasmin; fibroblasts exhibit increased accumulation of intracellular copper
GM3659A	5	EEMEM 20U	0078	24	F	WH	+-(D)	A	MOTHER	390	2	Two previous first trimester spontaneous abortions; son is only affected individual in a large pedigree; fibroblasts exhibit increased accumulation of intracellular copper
Wilson Disease (Hepatolenticular Degeneration) - 27790												
*GM0032	4	EEMEM 20U	0095	9	F	WH	--		PROBAND			Asymptomatic, patient on penicillamine; fibroblasts show elevated intracellular copper concentration
*GM0033	4	EEMEM 20U	0128	16	M	WH	--		PROBAND			Fibroblasts show elevated intracellular copper concentration
GM4128	3	EEMEM 20U	0107	20	F	WH	--		PROBAND			Negative family history; parents both have normal ceruloplasmin levels in serum; clinically affected
GM5257	2	EEMEM 20U	0409	34	F	WH	--		PROBAND	841	1	Diagnosed at age 24 as asymptomatic with "emotional instability"; low serum ceruloplasmin & high hepatic copper level; maintained on d-penicillamine; father had Wilson's disease; parents are first cousins; see GM5258 Lymphoid
GM5255	2	EEMEM 20U	0409	59	F	WH	+-(D)		MOTHER	841	2	Clinically unaffected; two affected children; see GM5256 Lymphoid
GM5259	3	EEMEM 20U	0409	30	F	WH	--		AFF SIS	841	3	Diagnosed at age 19 with "hepatitis"; maintained on d-penicillamine with resolution of liver disease & no evidence of CNS dysfunction; see GM5260 Lymphoid

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF METAL METABOLISM											
Wilson Disease (Hepatolenticular Degeneration) - 27790											
GM5339	2	EEMEM 20U	0409	25	F	WH	--	PROBAND	848	1	Onset at age 13 with jaundice, evidence of chronic liver disease, presence of Kayser-Fleischer rings, & low ceruloplasmin; controlled with d-penicillamine with no evidence of liver or CNS dysfunction; see GM5340 Lymphoid
GM5341	2	EEMEM 20U	0409	23	F	WH	--	AFF SIS	848	2	Diagnosed as asymptomatic at age 12 with low serum ceruloplasmin, elevated urinary & hepatic copper levels; maintained on d-penicillamine with no evidence of liver disease or CNS dysfunction; see GM5342 Lymphoid
GM5762	1	EEMEM 20U	0409	53	F	WH	+-(0)	MOTHER	848	3	No evidence of Wilson's disease; rheumatoid arthritis since age 21; two affected daughters; see GM5761A Lymphoid
GM5366A	5	EEMEM 20U	0409	32	F	WH	--	PROBAND	850	1	First diagnosed at age 25; low ceruloplasmin, hypersplenism, post-necrotic cirrhosis with portal hypertension & fluid retention, & elevated urinary copper excretion; on penicillamine; see GM5367 Lymphoid
GM5368	2	EEMEM 20U	0409	67	F	WH	+-(0)	MOTHER	850	2	Clinically unaffected
GM5370	2	EEMEM 20U	0409	70	M	WH	+-(0)	FATHER	850	3	See GM5371 Lymphoid; clinically unaffected
GM5798	2	EEMEM 20U	0409	37	F	WH	--	PROBAND	865	1	Diagnosed at age 25; neurological abnormalities; Kayser-Fleischer rings; low serum ceruloplasmin; inc. urinary copper excretion; elev. hepatic copper level; no clinical features of liver disease; on penicillamine; see GM5797 Lymphoid
GM5794	2	EEMEM 20U	0409	75	M	WH	+-(0)	FATHER	865	2	History of arthritis and coronary artery disease; see GM5793 Lymphoid
GM5796	4	EEMEM 20U	0409	70	F	WH	+-(0)	MOTHER	865	3	See GM5795 Lymphoid; clinically unaffected
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Adenine Phosphoribosyltransferase Deficiency - 10260											
*GM0517		EEMEM 20U	0071	64	M	BL	+-	PROBAND			Enzyme deficient in erythrocytes; normal level in fibroblasts; passage 3 at IMR

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency With Immunodeficiency - 10270												
*GM0469	8	EEMEM 20U	0118	1	M	WH	--	A	PROBAND			About 1% of normal ADA activity
*GM0471	4	EEMEM 20U	0103	18MO	F	BL	--	B	PROBAND			Two male sibs are similarly affected; no detectable RBC ADA
GM1390	5	EEMEM 20U	0090	3MO	M	WH	--	A	PROBAND			Short-limbed dwarfism; enzyme phenotypes: G6PD=B, Peptidases A, C, & D=1; deficient ADA activity
*GM2444	14	EEMEM 20U	0208	8	F	WH	--	A	PROBAND			French-Canadian; deficient ADA activity in RBC and fibroblasts; similarly affected male sib
*GM2446	6	EEMEM 20U	0208		F	WH	--	A	PROBAND			Deceased; Spanish-American; deficient ADA activity
GM3136		EEMEM 20U	0237	8MO	F	WH	--	A	PROBAND			Passage 2 at IMR; Sephardic Jew; classical early onset severe form; RBCs, fibroblasts, & mononuclear cells show ADA deficiency; 46,XX
GM3137		EEMEM 20J	0237		F		--	A	PROBAND			Passage 2 at IMR; classical early onset severe form; deficient ADA activity in erythrocytes, lymphocytes & fibroblasts; 46,XX/46,XX,t(1;12)(p36;q15); 52%/48%
*GM2027	9	EEMEM 20U	0118		F		+-(O)	B	PARENT	202	2	Mother of ADA deficient child; clinically normal; 53% of normal ADA activity in fibroblasts; proband not in Repository
*GM2028A	11	EEMEM 20U	0118		M		+-(O)	B	PARENT	202	3	Father of ADA deficient child; clinically normal; 48% of normal ADA activity in fibroblasts; proband not in Repository
*GM2434	6	EEMEM 20U	0208	7MO	M	WH	--	A	PROBAND	219	1	Deficient RBC and fibroblast ADA activity; similarly affected female sib; enzyme phenotypes: G6PD=B, 6PGD=A; peptidases A, C, & D=1; & PGM 1+3=1; see GM2471 Lymphoid
*GM2433	6	EEMEM 20U	0208	25	F	WH	+-(O)	A	MOTHER	219	2	Clinically normal; carrier level of ADA activity in RBCs, lymphocytes, and fibroblasts; see GM2472A Lymphoid
*GM2605	2	EEMEM 20U	0223	8MO	M	WH	--	A	PROBAND	226	1	See GM2606 Lymphoid; deficient fibroblast ADA activity
GM2607	3	EEMEM 20U	0223		F	WH	+-(O)	A	MOTHER	226	2	See GM2608 Lymphoid; carrier level of fibroblast ADA activity

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Adenosine Deaminase Deficiency With Immunodeficiency - 10270												
GM2609	2	EEMEM 20U	0223		M	WH	+-(0)	A	FATHER	226	3	See GM2610 Lymphoid; carrier level of fibroblast ADA activity
*GM2578	2	EEMEM 20U	0223	5MO	M	WH	--	A	PROBAND	232	1	Approximately 1% of normal ADA activity in RBCs, lymphocytes, and fibroblasts
GM2580	4	EEMEM 20U	0223	38	M	WH	+-(0)	A	FATHER	232	2	See GM2581A Lymphoid; carrier level of ADA activity in fibroblasts
GM2582	3	EEMEM 20U	0223	37	F	WH	+-(0)	A	MOTHER	232	3	See GM2583 Lymphoid; carrier level of ADA activity in fibroblasts
*GM2436	5	EEMEM 20U	0208	NB	F	WH	--	A	PROBAND	388	1	Swiss; deceased; deficient ADA activity in fibroblasts
GM2435	5	EEMEM 20U	0208	25	F	WH	+-(0)	A	MOTHER	388	2	Swiss; clinically normal; carrier level of fibroblast ADA activity
GM2447	11	EEMEM 20U	0208	25	M	WH	+-(0)	A	FATHER	388	3	Swiss; clinically normal; carrier level of fibroblast ADA activity
*GM2445	8	EEMEM 20U	0217	3	M	BL	--	A	PROBAND	423	1	See GM1715 Lymphoid; deficient ADA activity in fibroblasts; G6PD=A; GLO1=2; PGM1=1; PGM3=1; EST D=1; PGD=A; ME 2=1; AK 1=1; 46,XY; patient's HLA type is A2, 30,B7,39
GM2824	3	EEMEM 20U	0291	4MO	F	WH	--	A	PROBAND	446	1	See GM2825A Lymphoid; deficient ADA activity in RBCs, lymphocytes, and fibroblasts
Adenosine Deaminase Deficiency With No Immunodeficiency												
*GM1997	7	EEMEM 20U	0208	4MO	M	BL	--	A	PROBAND	633	1	Normal serum levels of IgG, IgM, IgA, & IgE; deficient RBC ADA activity; partial ADA deficiency in fibroblasts & lymphoblasts; see GM2294 Lymphoid
*GM3029	5	EEMEM 20U	0170	16	M	BL	--	B	PROBAND	634	1	!Kung South African; see GM3043 Lymphoid; clinically normal; partial ADA deficiency
*GM4395	2	EEMEM 20U	0251	6MO	M	WH	--	A	PROBAND	717	1	No detectable ADA activity in red cells or lymphocytes; see GM4396 Lymphoid; mother and brother have heterozygous levels of ADA activity; 14% normal ADA activity and cross immunoreactive protein in fibroblasts

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency - 14752												
GM1617	2	EEMEM 20U	0177	29	F	WH	+-		PROBAND	635	1	See GM1619 Lymphoid; deficient RBC ITPase
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
GM0068	14	EEMEM 10U	0027	2	M		Y-	A	PROBAND			Fibroblasts nonviable in HAT medium
*GM0152	10	EEMEM 20U	0101		M	WH	Y-	A	PROBAND			Fibroblasts nonviable in HAT medium
GM0537	7	EEMEM 20U	0031	13	M		Y-	A	PROBAND			Fibroblasts nonviable in HAT medium
*GM1362A	6	EEMEM 20U	0066	10	M	WH	Y-	A	PROBAND			Fibroblasts nonviable in HAT medium
*GM2330	9	EEMEM 10U	0039	20	M	WH	Y-	B	PROBAND			Variant; normal intelligence; no self-mutilation; HPRT activity in fibroblasts is reduced but not absent; cells survive in HAT medium
*GM3467	10	EEMEM 20U	0331	5DA	M	WH	Y-	B	PROBAND			Fibroblasts resistant to azaguanine & thioguanine; cannot utilize exogenous hypoxanthine; HPRT deficient
*GM0377A	11	EEMEM 20U	0095	8MO	M	WH	Y-	A	PROBAND	69	1	No detectable HPRT activity; formerly GM0029
*GM0013A	10	EEMEM 20U	0095	34	F	WH	+-(O)	A	MOTHER	69	2	46,XX; 55% of normal HPRT activity in fibroblasts
*GM0135	8	EEMEM 20U	0101		F	BL	+-(O)	A	PARENT	71	2	Sister of GM0318; HPRT+, G6PDB / HPRT-, G6PDA; 46,XX; mother of an HPRT deficient child; proband not in Repository; three affected nephews
*GM0318	13	EEMEM 20U	0101	40	F	BL	+-(O)	A	MAT AUNT	71	3	HPRT+, G6PDB / HPRT-, G6PDA; formerly GM0134; three affected sons & one affected nephew
*GM0159	11	EEMEM 20U	0085	10	M	BL	Y-	A	AFF MAT 1COU	71	4	G6PD type A; HPRT deficient; son of GM0318; two affected brothers & one affected maternal cousin
*GM2063	10	EEMEM 10U	0101	5	M	BL	Y-	A	AFF MAT 1COU	71	6	G6PD type A; see also GM0847 (SV40 Transformed); formerly GM0177; HPRT deficient; son of GM0318; two affected brothers & one affected maternal cousin
*GM1662	2	EEMEM 20U	0066	9	M	WH	Y-	A	PROBAND	72	1	No detectable HPRT, normal APRT activity
*GM1659	3	EEMEM 20U	0066	42	F	WH	+-(O)		MOTHER	72	2	Normal blood HPRT; carrier by hair root assay

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

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DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
GM1661	3	EEMEM 20U	0066	14	F	WH	+-		SIS	72	3	Normal blood HPRT; carrier by hair root assay
GM1660	3	EEMEM 20U	0066	11	F	WH	+-		SIS	72	4	Normal blood HPRT; carrier by hair root assay
*GM2227	3	EEMEM 20U	0066	12	M		Y-	A	PROBAND	73	1	No detectable HPRT; normal APRT activity
*GM2226	3	EEMEM 20U	0066	40	F		+-(D)	A	MOTHER	73	2	Two populations of cells: one showing no significant uptake and the other showing normal uptake of tritiated hypoxanthine by autoradiography; HPRT+ / HPRT-
*GM2290	2	EEMEM 20I	0183	19FW	M	WH	Y-	A	PROBAND	74	1	Fetal skin fibroblast culture; see also GM2292A Lymphoid and GM2338 Amniotic; same patient as GM2291; HPRT deficient; positive family history
*GM2291	2	EEMEM 20U	0183	19FW	M	WH	Y-	A	PROBAND	74	1	Fetal lung fibroblast culture; see also GM2292A Lymphoid and GM2338 Amniotic; same patient as GM2290; HPRT deficient; positive family history
*GM3116	2	EEMEM 20U	0183	19FW	M	WH	Y-	B	AFF BRO	74	2	Fetal kidney fibroblast culture; same fetus as GM3117
*GM3117	1	EEMEM 20U	0183	19FW	M	WH	Y-	B	AFF BRO	74	2	Fetal lung fibroblast culture; same fetus as GM3116
GM2851	3	EEMEM 20U	0066	15	M		Y-	A	PROBAND	508	1	Same mother as GM2852; deficient HPRT; normal APRT activity
GM2852	2	EEMEM 20U	0066	7	M		Y-	A	HALF BRO	508	2	Deficient HPRT; normal APRT activity
Myo-adenylate Deaminase Deficiency - 25475												
GM3071	4	EEMEM 20U	0178	14	M	WH	--		PROBAND	444	1	Skin fibroblast culture; muscle aching in all 4 extremities; elevated serum CPK; similarly affected 1st cousin; no detectable muscle ADA activity
GM3072	6	EEMEM 20U	0178	14	M	WH	--		PROBAND	444	1	Muscle fibroblast culture; same patient as GM3071
GM3073	5	EEMEM 20U	0178	38	F	WH	+-(D)		MOTHER	444	2	Skin fibroblast culture; 40% of normal muscle ADA activity
GM3074	6	EEMEM 20U	0178	38	F	WH	+-(D)		MOTHER	444	2	Muscle fibroblast culture; same patient as GM3073

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				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM								
Myo-adenylate Deaminase Deficiency - 25475												
GM3075	5	EEMEM 20U	0178	58	M	WH	+-(O)		FATHER	444	3	Skin fibroblast culture; 70% of normal muscle ADA activity
GM3076	4	EEMEM 20U	0178	58	M	WH	+-(O)		FATHER	444	3	Muscle fibroblast culture; same patient as GM3075
Orotic Aciduria - 25890												
*GM0328		EEMEM 20U	0080	2	M	WH	--	A	PROBAND			Line AUC; passage 2 at IMR; culture is moderately growth inhibited when medium lacks uridine, more severely inhibited when adenosine is added to uridine-deficient medium
*GM0632	2	EEMEM 20U	0115	14	F	WH	--	A	PROBAND			Culture is moderately growth inhibited when medium lacks uridine; more severely inhibited when adenosine is added to uridine-deficient medium
*GM1406	2	EEMEM 20U	0257		F	WH	--	A	PROBAND			Orotic acid crystals in urine; megaloblastic anemia; normal B12 & folic acid levels; pt. responded to oral uridine; cult is moder. growth inhib. in uridine def medium; severe growth inhib when adenosine is added to uridine-def medium
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM0544B	5	EEMEM 20U	0023	10	M	WH	--	A	PROBAND			De Sanctis-Cacchione; XP4LO; 2% of normal UV induced unscheduled DNA synthesis
GM2033	4	EEMEM 20U	0170	16	M	BL	--	A	PROBAND			XP3JO; Zulu; clinically affected
*GM2062	6	EEMEM 20U	0133	3	M	WH	--	A	PROBAND			XP5PJ; 46,XY; clinically affected
*GM2994	3	EEMEM 20U	0023	4	M	WH	--	A	PROBAND			Egyptian; XP5CA; onset of skin symptoms at age 1; photophobia; microcephaly and mental deficiency probably from birth; no skin or tongue neoplasia
GM0082	5	EEMEM 20U	0044	12	F	WH	--	A	PROBAND	76	1	Classical X.P. symptoms
GM0241	11	EEMEM 20U	0044		F	WH	+-(O)		MOTHER	76	2	Clinically unaffected
*GM1630	2	EEMEM 20U	0173	42MO	M	WH	--	A	PROBAND	77	1	XP1WI; small for age; speech problems; deficient repair activity; see GM2344 Lymphoid
*GM1631	2	EEMEM 20U	0173	29	M	WH	+-(O)	A	FATHER	77	2	Near normal repair; clinically normal; see GM7689 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group A - 27870												
GM1632	3	EEMEM 20U	0173	25	F	WH	+-(0)	A	MOTHER	77	3	Near normal repair; clinically normal; see GM7690 Lymphoid
GM2009	5	EEMEM 10U	0056	10	F	WH	--	A	PROBAND	200	1	XP1PD; mental and growth retardation; photosensitivity; similarly affected sib
GM2010	7	EEMEM 10U	0252	4	M	WH	--		AFF BRO	200	2	XP2PD; mental and growth retardation; photosensitivity
*GM2090	2	EEMEM 20U	0170	16	M	BL	--	A	PROBAND	389	1	XP4JO; African; positive family history; blindness due to corneal opacity; skin shows areas of hypo- and hyperpigmentation but no malignancies; offspring of consanguineous parents
GM2091	3	EEMEM 20U	0170	31	M	BL	--	A	AFF 3RD COU	389	2	XP5JO; African; blindness due to corneal opacity; skin shows areas of hypo- and hyperpigmentation but no malignancies; offspring of consanguineous parents
*GM2990	2	EEMEM 20U	0023	8	F	WH	--	A	PROBAND	468	1	Egyptian; XP1CA; parents are 1st cousins; onset of skin symptoms at age 1; photophobia; microcephaly and mental deficiency probably from birth; one similarly affected sib; hypersensitive to UV induced cell killing & mutagenesis
*GM2991	3	EEMEM 20U	0023	6	M	WH	--	A	AFF 1ST COU	468	2	Egyptian; XP2CA; parents are 1st cousins; onset of skin symptoms at age 1; photophobia; microcephaly and mental deficiency probably from birth; two similarly affected sibs
*GM5509	2	EEMEM 20U	0226	17	F	WH	--	B	PROBAND	484	1	XP12BE; multiple skin cancers; acute sun sensitivity; nervous system involvement; see GM2250A Lymphoid and GM4429B SV40 transformed fibroblast; less than 2% of normal UV induced unscheduled DNA synthesis
GM5510	2	EEMEM 20U	0226	44	F	WH	+-(0)		MOTHER	484	2	Clinically normal; see GM5511A Lymphoid
GM5566	2	EEMEM 20U	0226	20	M	WH	+?		BRO	484	3	Clinically normal; possible heterozygote; see GM5567 Lymphoid
GM5568	3	EEMEM 20U	0226	47	M	WH	+-(0)		FATHER	484	4	Clinically normal; see GM5569A Lymphoid
*GM4314A		EEMEM 20U	0335	5	F	OR	--	B	AFF SIS	695	2	Japanese; XP3OS; severe XP; mental retardation; gait disturbance; less than 2% of normal post UV unscheduled DNA synthesis; passage 5 at IMR; slow grow-

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM0710A	16	EEMEM 20U	0011	18MO	M	WH	--	B	PROBAND	703	1	De Sanctis-Cacchione; XP25RO; ATCC CRL 1261; 0 to 5% of normal post UV light induced unscheduled DNA synthesis
GM5292	8	EEMEM 20U	0335	8	F	OR	--	B	PROBAND	842	1	Japanese; XP3KR; dry, pigmented skin; mental retardation; similarly affected identical twin; less than 5% of normal post UV light induced unscheduled DNA synthesis
Xeroderma Pigmentosum: Complementation Group B - 27871												
*GM1854	3	EEMEM 20U	0155	64	F	WH	+-(O)		MOTHER	283	2	Mother of GM2252A Lymphoid; see also GM1855 Lymphoid; also heterozygous for Cockayne syndrome
Xeroderma Pigmentosum: Complementation Group C - 27872												
*GM0510	9	EEMEM 10U	0023	10	M	WH	--	A	PROBAND			XP1PW; Arab; low post UV light induced unscheduled DNA synthesis
*GM0709	17	EEMEM 20U	0011	15	M	WH	--	B	PROBAND			XP21RO; Dutch; 10 to 15% of normal post UV light induced unscheduled DNA synthesis
*GM2992	3	EEMEM 20U	0023	13	F	WH	--	A	PROBAND			Egyptian; XP3CA; onset of skin symptoms at age 3; photophobia; with no CNS involvement; one similarly affected sib
*GM2997	3	EEMEM 20U	0023	4	M	WH	--	A	PROBAND			Egyptian; consanguineous parents; XP7CA; onset of skin symptoms at age 9 mo; photophobia; no CNS involvement or skin neoplasia
GM5293	9	EEMEM 20U	0335	6	F	OR	--	B	PROBAND			Japanese; XP2KA; dry, pigmented skin; skin cancer; no mental retardation; 25% of normal post UV light induced unscheduled DNA synthesis
*GM0671	4	EEMEM 20U	0023	15	M	WH	--	A	PROBAND	75	1	XP8BE; ATCC CRL 1158; 10-20% of normal UV induced unscheduled DNA synthesis; see GM2249B Lymphoid; similarly affected twin and two brothers
GM0676	3	EEMEM 20U	0023	15	M	WH	--		AFF TWIN	75	2	XP9BE; ATCC CRL 1161; 10-20% of normal UV induced unscheduled DNA synthesis; see GM2498 Lymphoid; similarly affected twin and two brothers
GM0673	4	EEMEM 20U	0023	21	M	WH	--		AFF BRO	75	3	XP4SL; three affected brothers

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group C - 27872												
*GM0677	3	EEMEM 20U	0023	25	M	WH	--	A	AFF BRO	75	4	XP2BE; ATCC CRL 1166; 19% of normal UV induced unscheduled DNA synthesis; three affected brothers
GM2032	2	EEMEM 20U	0170	5	M	BL	--	B	PROBAND	78	1	XP2JO; Zulu; tongue tumor fibroblast culture; same patient as GM2035; similarly affected sib
*GM2035	3	EEMEM 20U	0170	5	M	BL	--	A	PROBAND	78	1	XP2JO; Zulu; skin fibroblast culture; same patient as GM2032; similarly affected sib
*GM2034	4	EEMEM 20U	0170	28	F	BL	+-(0)		MOTHER	78	2	XPH2JO; Zulu; two affected sons; clinically normal
*GM2024	2	EEMEM 20U	0170	3	M	BL	--		AFF BRO	78	3	XP1JO; Zulu; several carcinomas of the oral cavity
GM1736	2	EEMEM 20U	0133	21	F	WH	--	A	PROBAND	415	1	Diagnosed at age 3 yrs
GM3080	6	EEMEM 20U	0133	59	M	WH	+-(0)		FATHER	415	2	Clinically normal
GM3081A	6	EEMEM 20U	0133	51	F	WH	+-(0)		MOTHER	415	3	Clinically normal
*GM2993	3	EEMEM 20U	0023	8	F	WH	--	A	PROBAND	466	1	Egyptian; XP4CA; consanguineous parents; onset of skin symptoms at age 3-4; photophobia; no CNS involvement; with both tongue and skin neoplasia
*GM2998	2	EEMEM 20U	0023		F	WH	+-(0)		MOTHER	466	2	Egyptian; XPH4CA; clinically normal
*GM2995	3	EEMEM 20U	0023	10	M	WH	--	A	PROBAND	467	1	Egyptian; XP6CA; onset of skin symptoms at age 11 mo; photophobia; no CNS involvement; leukoplakia and skin neoplasia; one similarly affected sib
*GM2996	2	EEMEM 20U	0023	7WK	M	WH	--	A	AFF BRO	467	2	Egyptian; XP8CA; no clinical symptoms of XP at time of biopsy but fibroblasts show defective DNA repair; onset of skin symptoms at age 4 mo; no photophobia, CNS involvement, or skin neoplasia; one similarly affected sib
*GM0522	6	EEMEM 20U	0037	16	F	BL	--	A	PROBAND	503	1	G6PD type B; Phosphoglucomutase 1; XP2NB1; similarly affected sib
*GM0523A	9	EEMEM 20U	0037	10	F	BL	--	A	AFF SIS	503	2	G6PD type B; Phosphoglucomutase 1; XP1NB1

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMNS	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group C - 27872												
*GM0030A	8	EEMEM 20U	0110	24	M	WH	--	B	PROBAND	507	1	XP3BE; ATCC CRL 1189; 10-20% of normal UV induced unscheduled DNA synthesis; see GM2248A Lymphoid
*GM3176	3	EEMEM 20U	0023	3	F	WH	--	A	PROBAND	600	1	XP1AA; 10-20% of normal UV induced unscheduled DNA synthesis
GM3177	2	EEMEM 20U	0023	2	M	WH	+?	A	BRO	600	2	Clinically unaffected; possible heterozygote; normal level of UV induced unscheduled DNA synthesis
GM3178	2	EEMEM 20U	0023	30	F	WH	+-(D)	A	MOTHER	600	3	XPH1AA; Asian; normal level of UV induced unscheduled DNA synthesis; clinically normal
GM3179	2	EEMEM 20U	0023	32	M	WH	+-(D)	A	FATHER	600	4	XPH2AA; Asian; normal level of UV induced unscheduled DNA synthesis; clinically normal
*GM2096	3	EEMEM 20U	0007	15	F	BL	--	A	PROBAND	627	1	De Sanctis-Cacchione; lupus erythematosus; see GM2634 Lymphoid; XP1MI; 46,XX; low level of UV induced unscheduled DNA synthesis
Xeroderma Pigmentosum: Complementation Group D - 27873												
GM0436	3	EEMEM 20U	0023	22	F	WH	--	A	PROBAND			XP1NE; near normal post UV unscheduled DNA synthesis; no neoplasias
*GM0936	8	EEMEM 20U	0046	22FW	F	WH	--	A	PROBAND			Umbilical cord fibroblast culture; low in post UV unscheduled DNA synthesis
GM1295	4	EEMEM 20U	0145	6	F	WH	--	A	PROBAND			XP1KC; extreme photosensitivity, photophobia, and rhinorrhea; negative family history
*GM3615	11	EEMEM 20U	0023		F		--	A	PROBAND			XP1BR; clinically affected
*GM5424	3	EEMEM 20U	0023	2	M	WH	--	A	PROBAND			XP1PO; erythema, freckling, scaling, & drying of skin; neurological problems; imperforate anus, & peripheral pulmonic stenosis; paternal side of family has a history of Charcot-Marie-Tooth disease; 46,XY
*GM0434	3	EEMEM 20U	0023	28	M	WH	--	A	PROBAND	502	1	De Sanctis-Cacchione; XP3NE; 30 to 50% of normal post UV unscheduled DNA synthesis; similarly affected sib

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Xeroderma Pigmentosum: Complementation Group D - 27873											
*GM0435	3	EEMEM 20U	0023	23	F	WH	-- A	AFF SIS	502	2	De Sanctis-Cacchione; XP2NE; 30 to 50% of normal post UV unscheduled DNA synthesis
Xeroderma Pigmentosum: Complementation Group E - 27874											
*GM2415	12	EEMEM 20U	0011	34	F	WH	-- B	PROBAND	626	1	XP2RO; ATCC CRL 1259; 2nd cousin of XP3RO (GM2450C Lymphoid); clinically affected; increased incidence of chromatid gaps following x-irradiation
Xeroderma Pigmentosum: Complementation Group F - 27876											
*GM3542	9	EEMEM 20U	0335	29	M	OR	-- B	PROBAND			Japanese; XP3YO; 10% of normal post UV unscheduled DNA synthesis; no neurological abnormalities
GM4313A	15	EEMEM 20U	0335	64	F	OR	-- B	PROBAND			Japanese; XP2YO; mild symptoms of XP; skin cancers; 10% of normal post UV unscheduled DNA synthesis
Xeroderma Pigmentosum: Complementation Group G - 27878											
*GM3021A	13	EEMEM 20U	0011	17	F		-- B	PROBAND			XP2BI; neurological abnormalities clinically well manifested at age 11; acute sun sensitivity; fibroblasts show no unscheduled DNA synthesis following exposure to UV irradiation
*GM3616	10	EEMEM 20U	0023		M		--	PROBAND			XP3BR; clinically affected
Xeroderma Pigmentosum: Complementation Group M											
GM3248	2	EEMEM 20U	0110	5	M	WH	-- A	PROBAND	714	1	Also possible Cockayne Syndrome; 46,XY; 10% of cells show random chromosome loss/gain; XP skin changes in areas exposed to light; neurological manifestations; see GM3249 Lymphoid
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878											
*GM1213	2	EEMEM 20U	0133	61	F	WH	-- A	PROBAND			Diagnosed at age 34; more resistant to UV light than other XP cell lines; high DNA repair capacity; complements groups A, C, and D; possible candidate for group E
*GM1227	3	EEMEM 20U	0133	28	M	WH	-- A	PROBAND			Diagnosed at age 13; 30-60% of normal post UV light induced unscheduled DNA synthesis with normal strand incision
GM1509	3	EEMEM 20U	0133	38	F	WH	--	PROBAND			Clinically affected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878												
GM2476	2	EEMEM 20U	0170	10	F	BL	--		PROBAND			Zulu; XP6JO; no neurological abnormalities; no mental retardation
GM3083	6	EEMEM 20U	0133	54	F	WH	+-(O)		PARENT			Mother of an affected child; proband not in Repository
GM3614	3	EEMEM 20U	0110	21	F	BL	--		PROBAND	495	1	Microcephaly, retarded growth, mental deficiency; sickle cell trait; hypo- and hyperpigmented skin areas
Xeroderma Pigmentosum: Variants - 27875												
*GM2004	3	EEMEM 20U	0023	30	F	WH	--	A	PROBAND			Patient shows gradual mental retardation; XP1WA; 46,XX; 50% of cells have unidentified 2q+
*GM3617	11	EEMEM 20U	0023	30	M		--	A	PROBAND			XP30RO; variant; light to moderate XP symptoms; 88% of control level of unscheduled DNA synthesis with normal strand incision
*GM3618	7	EEMEM 20U	0023		F		--	A	PROBAND			XP6OU; variant; clinically affected
GM6090	5	EEMEM 20U	0023	42	F	OR	--	B	PROBAND			Chinese; XP1SF; skin hyperpigmentation and solar cancers on face; one similarly affected sib; abnormal DNA replication after UV irradiation
*GM3379	2	EEMEM 20U	0323	86	F	WH	--	A	PROBAND	220	1	XP5HA; "pigmented xerodermoid"; XP variant; apparently normal exc'sion repair & a hypersensitive response of semiconservative DNA replication to UV damage; three similarly affected sibs
GM1389	4	EEMEM 20U	0133	21	F	WH	--	A	PROBAND	377	1	See GM1646A Lymphoid; diagnosed at age 16 yrs; fibroblasts show normal UV sensitivity and repair but variant DNA replication post UV irradiation
*GM2359	3	EEMEM 20U	0046	25	M	WH	--	A	PROBAND	433	1	Variant; DNA excision repair normal; DNA post-replication repair is abnormal; Iranian; XP115LO; same patient as GM2111; parents are first cousins
*GM3055	6	EEMEM 20U	0023	28	M	WH	--	A	PROBAND	438	1	XP1CH; 55% of normal unscheduled DNA synthesis with normal strand incision; parents are first cousins; similarly affected sib

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Variants - 27875												
*GM3053	10	EEMEM 20U	0023	23	F	WH	--	A	AFF SIS	438	2	XP2CH; 56% of normal post UV light induced unscheduled DNA synthesis with normal strand incision; parents are 1st cousins; similarly affected sib
*GM3054	9	EEMEM 20U	0023	50	F	WH	+-(0)	A	MOTHER	438	3	XPH1CH; Russian-Armenian; normal unscheduled DNA synthesis and repair replication; two affected children
Sun Sensitive High Cancer Syndrome												
*GM2881	8	EEMEM 20U	0294	67	M	WH			PROBAND			First skin tumor at age 34; 4 of 4 sibs affected as well as 2 nephews; no abnormality of DNA repair or replication in fibroblasts; pedigree shows dominant inheritance
DISORDERS OF STEROID METABOLISM												
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191												
GM2241	2	EEMEM 20U	0107	1MO	F	WH	--		PROBAND	85	1	Genital tissue; see GM2242 Lymphoid; 21-hydroxylase deficiency; salt-losing type; responsive to hydrocortisone treatment
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010												
GM4496	4	EEMEM 20U	0178	6	M	WH	Y-	B	PROBAND			High proportion of fatty acids with a chain length of 26 found in fibroblasts; autopsy pathological findings in both the adrenal and central nervous system were those of ALD
GM4932	5	EEMEM 20U	0175	10	M	BR	Y-	B	PROBAND			Maori tribe, New Zealand; many affected individuals in pedigree; elevated C26:C22 fatty acid ratio in fibroblasts; no clinical evidence of adrenal insufficiency; only microscopic evidence observed at autopsy
*GM4933	6	EEMEM 20U	0175	8	M	WH	Y-	B	PROBAND			Affected uncles and cousins; elevated C26:C22 fatty acid ratio in fibroblasts and plasma; onset at age 2; normal neurological examination
GM4934	5	EEMEM 20U	0175	7	M	WH	Y-	B	PROBAND			Elevated C26:C22 fatty acid ratio in fibroblasts and plasma
GM7530	6	EEMEM 20U	0175	26	M	WH	Y-	B	PROBAND			Adrenomyeloneuropathy; adrenal insuff.; progress. spastic paraparesis; occasional nausea & vomiting; increased freq. of urination; elevated levels of C26 fatty acids in plasma & fibro.; neg. family history

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF STEROID METABOLISM												
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010												
*GM7531	6	EEMEM 20U	0175	19	M	WH	Y-	B	PROBAND			Adrenomyeloneuropathy; adrenal insuff. since age 11; unsteady gait since age 13; sl. wkness & hyperreflexia since age 19; bro. has mild spastic paraparesis; moth. had progressive paraparesis; elev. level of C26 fatty acid in plasma & fibr
*GM7675	5	EEMEM 20U	0175	22	M	WH	Y-	B	PROBAND			Adrenomyeloneuropathy; neurologically norm. until age 20; urinary tract dysfunction & mild spasticity in legs since age 22; abnorm. EEG with posterior slowing; elev. levels of C26 fatty acids in plasma & fibro.; positive family history
GM0337	3	EEMEM 20U	0105	35	F	WH	+?		PARENT	86	2	Mother of an affected child; proband not in Repository
GM4904	7	EEMEM 20U	0175	11	M	WH	Y-	A	PROBAND	764	1	Elevated C26:C22 fatty acid in fibroblasts; adrenal insufficiency; progressive white matter disease; progressively deteriorating intellectual functioning; one 2nd & two 3rd maternal cousins have ALD; see GM4673 Lymphoid
GM4903	4	EEMEM 20U	0175	30	F	WH	+-(O)	A	MOTHER	764	2	Clinically normal; one first cousin and two second cousins have adrenoleukodystrophy; abnormal C26:C22 fatty acid ratio in plasma & cultured fibroblasts; see GM4674 Lymphoid
Primary Cortisol Resistance												
*GM4901	7	EEMEM 20U	0389	56	M	WH		B	PROBAND	766	1	Dutch; elevated plasma & urinary free cortisol without Cushing's disease; hypertension; hypokalemia; normal number but decreased affinity of fibroblast receptors for cytosol; positive family history
*GM4902	8	EEMEM 20U	0389	26	M	WH		B	AFF SON	766	2	Dutch; elevated plasma & urinary free cortisol without Cushing's disease; no clinical manifestations; normal number but decreased affinity of fibroblast receptors for cytosol
Male Pseudohermaphroditism: 17-Ketosteroid Reductase Deficiency - 26430												
GM3547	5	EEMEM 20U	0077	15	F	WH	--		PROBAND			Arab; 46,XY; ambiguous genitalia, virilized at puberty; similarly affected sister; parents are first cousins

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF STEROID METABOLISM												
Pseudovaginal Perineoscrotal Hypospadias; 5-Alpha-Reductase Def - 26460												
*GM2935	9	EEMEM 20U	0077	16	F	WH	--	B	PROBAND	461	1	Arab; skin biopsy (labialscrotal fold); similarly affected sister; parents are 1st cous.; elevated blood testosterone/dihydrotestosterone ratio; 46,XY in leukocytes & fibroblasts; presumed 5-alpha reductase deficiency
Testicular Feminization Syndrome (Androgen Insensitive/Receptor Def) - 31370												
GM1404	4	EEMEM 20U	0109	20	F	BL	Y-		PROBAND			46,XY; phenotypically female
GM2299	3	EEMEM 20U	0107	8MO	F	BL	Y-		PROBAND	123	1	46,XY; genital skin fibroblast culture; half-sister of GM2682; positive family history
GM2300	2	EEMEM 20U	0107	8MO	F	BL	Y-		PROBAND	123	1	46,XY; non-genital skin fibroblast culture; same patient as GM2299
GM2301	4	EEMEM 20U	0107	8MO	F	BL	Y-		PROBAND	123	1	46,XY; gonad fibroblast culture; same patient as GM2299
GM2682	4	EEMEM 20U	0107	7	F	BL	Y-		AFF SIS	123	2	Thigh skin; 46,XY; same patient as GM2683
GM2683	4	EEMEM 20U	0107	7	F	BL	Y-		AFF SIS	123	2	Genital skin; 46,XY; same patient as GM2682
GM2715	2	EEMEM 20U	0107	14	F	WH	Y-		PROBAND	244	1	Genital skin; 46,XY; no measurable dihydrotestosterone binding to fibroblasts
GM2716	3	EEMEM 20U	0107	14	F	WH	Y-		PROBAND	244	1	Epididymus tissue; 46,XY; same patient as GM2715
GM2717	2	EEMEM 20U	0107	14	F	WH	Y-		PROBAND	244	1	Non-genital skin; 46,XY; same patient as GM2715
GM1721	2	EEMEM 20U	0107	15	F	WH	Y-		PROBAND	393	1	46,XY; abdominal skin fibroblast culture; genital tissue fibroblast culture (GM1722) is also available
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Achondrogenesis; Type IA - 20060; Type IB - 20061; and Type II - 20070												
GM1892	6	EEMEM 20U	0417	32FW	F	WH	--		PROBAND			Ashkenazi; stillborn; marked limb shortening; macrocephaly; cleft palate; depressed nasal bridge; micrognathia; polyhydramnios by ultrasound; radiograph shows shortened long bones, decreased mineralization, norm. calvarium; Type IB

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Campomelic Dwarfism (Campomelic Dysplasia or Syndrome) - 21197												
GM1737	9	EEMEM 20U	0142	9DA	F	WH	--		PROBAND			Gonad biopsy; phenotypically female; 46, XY with random chromosome loss; normal fibroblast alkaline phosphatase activity
GM4329	2	EEMEM 20U	0107	6WK	F	WH	--		PROBAND			Multiple abnormalities of extremities including anterior bending of the femur and tibia; two maternal uncles have a history of congenital deformities of the lower limbs; normal fibroblast alkaline phosphatase activity; 46,XX
Cartilage-Hair Hypoplasia (Metaphyseal Chondrodysplasia) - 25025												
GM1671	4	EEMEM 20U	0070	17	M		--		PROBAND			Slightly elevated level of adenosine deaminase activity in fibroblasts
GM3625	8	EEMEM 20U	0134	38	F	WH	--		PROBAND			Amish; short stature; thin hair; no history of immune defect
GM3626	7	EEMEM 20U	0134	20	M	WH	--		PROBAND			Old order Amish; short stature, T cell defect, and thin hair
Central Core Disease of Muscle - 11700												
GM3728	3	EEMEM 20U	0348	53	M	WH	+-		PROBAND	529	1	Weakness & positive muscle biopsy - no type II fibers, type I fibers have central cores; positive family history; 46, XY/46,XY,t(7;11)(7pter>7q11::11p11> 11pter;11qter>11p11::7q11>7qter); balanced; 94%/6%
Chondrodystrophy												
GM0229	3	EEMEM 20U	0107	9MO	M	WH	-?		PROBAND			Mother also affected; 46,XY
Cockayne Syndrome - 21640												
*GM0739	3	EEMEM 20U	0115	3	F	WH	--	A	PROBAND			46,XX; increased UV light-induced sister chromatid exchanges and cell killing; patient is CS1AN; complementation group B
*GM1428	9	EEMEM 20U	0091	8	F	BL	--		PROBAND			Fibroblasts have markedly decreased post UV light colony-forming ability; patient is CS7SE; complementation group B
*GM1098B	7	EEMEM 20U	0006	20	M	WH	--		PROBAND	251	1	46,XY; see GM1712 Lymphoid; sensitive to UV irradiation; patient is CS2BE or CS5HO; complementation group B
*GM2856	4	EEMEM 20U	0155	13	M	WH	--	B	PROBAND	399	1	Fibroblasts have markedly decreased post UV light colony-forming ability; same x-ray sensitivity as normal cells; see GM1857 Lymphoid; pt. is CS3BE; complementation group A

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Cockayne Syndrome - 21640												
*GM1629	3	EEMEM 20U	0155	10	F	WH	--	B	PROBAND	628	1	Father is GM2474A Lymphoid; mildly retarded; small skull; spastic diplegia; fibroblasts have decreased post UV light and x-ray colony-forming ability; pt. is CS1BE; complementation group B
*GM2965	2	EEMEM 20U	0210	25	F	WH	--		PROBAND	636	1	Atypical; late age of onset; lack of ocular and retinal lesions; see GM2964 Lymphoid; complementation group A
*GM2838	3	EEMEM 20U	0264	24	M	WH	--		PROBAND	995	1	Profound dwarfing; typical facies; ataxic; premature aging and death; two similarly affected sibs
Conradi Syndrome (Chondrodysplasia Punctata) - 21510												
GM0740	2	EEMEM 20U	0107	1	F	WH	--		PROBAND			Clinically affected
Cornelia de Lange Syndrome (Brachmann-de Lange Syndrome) - 12247												
GM0045	5	EEMEM 20U	0066	13	M	WH	+-		PROBAND			46,XY; some chromosome loss and structural aberrations in 70% of fibroblasts
GM3478	2	EEMEM 20U	0107	4DA	M	WH	+-		PROBAND			Typical facies; clinodactyly; fibroblasts show 46,XY/92,XXYY; 76%/24% with fragments, breakage, and random loss
Cutis Laxa - 21910												
GM0480		EEMEM 20U	0066	45	F	WH	--		PROBAND			Passage 4 at IMR; clinically affected
GM1353	3	EEMEM 20U	0147	12DA	M	WH	--		PROBAND			46,X,Yp+; clinically affected
GM1377	4	EEMEM 20U	0147	8WK	M	BL	--		PROBAND			Redundant skin on arms and thighs; hyperextensible MP and IP joints
GM2739	2	EEMEM 20U	0232	17	M	WH	--		PROBAND			Classical symptoms
GM2768	5	EEMEM 20U	0147	4MO	F	WH	--		PROBAND			Clinically affected
Dyggve-Melchior-Clausen Disease - 22380												
GM2437	3	EEMEM 20U	0070		F		--		PROBAND			Clinically affected
*GM4997		EEMEM 20U	0394	23	M	WH	--		PROBAND			Lebanese; consanguineous parents; two affected sibs; mental deficiency and dwarfism; passage 3 at IMR
Dyschondrosteosis - 12730												
GM2885	2	EEMEM 20U	0315	47	F	WH	+-		PROBAND			Type of Leri and Weil
Dystonia Musculorum Deformans, Dominant Form (Torsion Dystonia) - 12810												
GM3211	2	EEMEM 20U	0307	30	M	WH	+-		PROBAND			Generalized dystonia; Ashkenazi; age of onset 7 yrs; mother and sister also affected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMC	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE											
Dystonia Musculorum Deformans; Dominant Form (Torsion Dystonia) - 12810											
GM3221	3	EEMEM 20U	0307	50	F	WH	+-	PROBAND			Focal dystonia; father and grandfather also affected; onset at age 43 years
GM2304	3	EEMEM 20U	0184	17	F	WH	+-	PROBAND	102	1	See GM2305 Lymphoid; onset at age 7; at time of biopsy very little speech and considerable dystonia of all extremities
Dystonia Musculorum Deformans; Recessive Form (Torsion Dystonia) - 22450											
GM3208	2	EEMEM 20U	0307	29	M	WH	--	PROBAND			Generalized dystonia; Ashkenazi; onset at age 10 years
GM3218	2	EEMEM 20U	0307	25	M	WH	--	PROBAND			Generalized dystonia; similarly affected brother; onset at age 2 years
*GM2306	2	EEMEM 20U	0184	13	M	WH	--	PROBAND	103	1	See GM2307 Lymphoid; onset at age 10; father is Jewish
*GM2255	3	EEMEM 20U	0184	14	F		--	PROBAND	473	1	See GM2256 Lymphoid; onset at age 12; severe involvement of trunk and extremities; similarly affected sister
*GM2551	3	EEMEM 20U	0184	31	F	WH	--	PROBAND	517	1	Eastern European Jewish; onset at age 15
Dystonia Musculorum Deformans; Type Unknown											
GM3209	3	EEMEM 20U	0307	49	F	WH		PROBAND			Focal (axial) dystonia; age of onset 41 yrs
GM3210	3	EEMEM 20U	0307	21	M	WH		PROBAND			Generalized dystonia; age of onset 2 yrs; negative family history
GM3213	2	EEMEM 20U	0307	26	F	WH		PROBAND			Generalized dystonia; age of onset 12 yrs
GM3214	3	EEMEM 20U	0307	37	M	WH		PROBAND			Focal (spasmodic torticollis) dystonia; age of onset 35 yrs; negative family history
GM3215	2	EEMEM 20U	0307	48	F	WH		PROBAND			Segmental dystonia; age of onset 44 yrs; negative family history
GM3216	2	EEMEM 20U	0307	64	F	WH		PROBAND			Focal (oral-mandibular) dystonia; age of onset 59 yrs; negative family history
GM3217	2	EEMEM 20U	0307	49	M	WH		PROBAND			Segmental dystonia; age of onset 41 yrs; negative family history
GM3219	4	EEMEM 20U	0307	18	M	WH		PROBAND			Segmental dystonia; Ashkenazi; age of onset 10 yrs; negative family history

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
				DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE								
Dystonia Musculorum Deformans: Type Unknown												
GM3220	4	EEMEM 20U	0307	35	M	WH			PROBAND			Focal (spasmodic torticollis) dystonia; age of onset 33 yrs; negative family history
GM3222	4	EEMEM 20U	0307	71	F	WH			PROBAND			Focal (spasmodic torticollis) dystonia; Ashkenazi; age of onset 67 yrs; negative family history
Ehlers-Danlos Syndrome: Type I (Severe Form; E-D Gravis) - 13000												
GM1812	3	EEMEM 20U	0171		F		+-		PROBAND			Clinically affected
GM2007	6	EEMEM 20U	0171	38	F	WH	+-		PROBAND			Clinically affected
GM2691	6	EEMEM 20U	0147	17	F	WH	+-		PROBAND			Clinically affected
Ehlers-Danlos Syndrome: Type II (E-D Mitis) - 13002												
GM1788	2	EEMEM 20U	0171	51	M	WH	+-		PROBAND			Clinically affected
GM1691	3	EEMEM 20U	0171	19	M	WH	+-		PROBAND	104	1	Same patient as GM2006; clinically affected
Ehlers-Danlos Syndrome: Type IV (Arterial, Ecchymotic Type) - 13005, 22535												
GM2022	7	EEMEM 10U	0147	18	F	WH			PROBAND	362	1	Recurrent aneurysms; blue sclerae; hypomobile thumbs; mode of inheritance uncertain; negative family history
Ehlers-Danlos Syndrome: Type VI - (Hydroxylysine-deficient Collagen) - 22540												
*GM1790	15	EEMEM 20U	0147	12	F	WH	--	A	PROBAND	105	1	Parents not affected; kyphoscoliosis; recurrent joint dislocation; hyperextensible skin & joints; decreased collagen hydroxylysine content; similarly affected sib; 30% of normal lysyl hydroxylase
*GM1791	15	EEMEM 20U	0147	9	F	WH	--	A	AFF SIS	105	2	Kyphoscoliosis; recurrent joint dislocation; hyperextensible skin and joints; decreased collagen hydroxylysine content; 30% of normal collagen lysyl hydroxylase activity
Ehlers-Danlos Syndrome: Type Unclassified												
GM0733	3	EEMEM 20U	0143	7	M	WH			PROBAND			Clinically affected
GM1161	3	EEMEM 20U	0095	8	F				PROBAND			Clinically affected
GM1897	3	EEMEM 20U	0147	32	F	WH			PROBAND			Clinically affected
GM2293	2	EEMEM 20U	0185	14	F	WH			PROBAND			Cuban; bilateral congenital glaucoma; loose skin; hyperpigmented skin; hypertrophic scars; father and brother are similarly affected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Ehlers-Danlos Syndrome: Type Unclassified												
GM2681	2	EEMEM 20U	0147	36	F	WH			PROBAND			Clinically affected
*GM3850	4	EEMEM 20U	0355	23	F	WH			PROBAND			Severe hernias; mild joint hypermobility; asthmatic bronchitis; eczematoid dermatitis; severe scoliosis; died from ruptured aorta; similarly affected son; possibly dominant inheritance
GM2206	3	EEMEM 20U	0147	17	F	WH		A	PROBAND	208	1	Variant; hydroxylysine deficient, not lysyl hydroxylase deficient; 46,XX, inv(6)(pter>p24::p21>p24::p21>qter)pat; formerly GM1514
GM2205	4	EEMEM 20U	0147	51	M	WH		A	FATHER	208	2	Clinically unaffected; 46,XY,inv(6)(pter>p24::p21>p24::p21>qter); formerly GM1515
GM1516	2	EEMEM 20U	0147	49	F	WH			MOTHER	208	3	Clinically unaffected; 46,XX
GM2490	9	EEMEM 10I	0147	11	F	WH			PROBAND	483	1	Joint laxity; thin skin; similarly affected sister
Epidermolysis Bullosa Dystrophica - Recessive Inheritance - 22660												
GM2857	2	EEMEM 20U	0232	12	M	WH	--		PROBAND			Puerto Rican; negative family history; clinically affected
Myopathy with Giant Abnormal Mitochondria - 25514												
GM0028	4	EEMEM 20U	0095	21	F	WH	--		PROBAND			Lebanese; 46,XX; skeletal muscle hypermetabolism; abnormal mitochondria; genetic etiology unproven; 2nd reported case
Hypophosphatasia (Phosphoethanolaminuria) - 24150												
*GM3459	3	EEMEM 20U	0330	5MO	M	WH	--	A	PROBAND			Severe lethal, in utero onset; deficient serum & fibroblast alkaline phosphatase activity
*GM3859	5	R1640 10U	0260	1DA	F	WH	--	A	PROBAND			Severe, in utero onset; deficient liver, bone, and fibroblast alkaline phosphatase activity
*GM1571	3	EEMEM 20U	0095	20FW	M	BL	--	A	PROBAND	371	1	Fetal skin; in utero onset; fatal; deficient fibroblast alkaline phosphatase activity; similarly affected sib
GM3191	3	EEMEM 20U	0078	30	M	WH	+-(D)		PARENT	509	1	Father of child with fatal, in utero onset form; proband not in Repository

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Hypophosphatasia (Phosphoethanolaminuria) - 24150												
GM3192	3	EEMEM 20U	0078	25	F	WH	+-(0)		PARENT	509	2	Mother of child with fatal, in utero onset form; proband not in Repository
GM3502		EEMEM 20U	0321	20FW	F	WH	--	A	PROBAND	604	1	See GM3336 Amniotic; severe, lethal, in utero onset; passage 2 at IMR; similarly affected sib; 46,XX; deficient fibroblast alkaline phosphatase activity
GM3328		EEMEM 20U	0321		F	WH	+-(0)		MOTHER	604	2	Passage 4 at IMR; two affected children
GM3327		EEMEM 20U	0321	25	M	WH	+-(0)		FATHER	604	3	Passage 4 at IMR; two affected children
*GM4235	6	EEMEM 20U	0321	5MO	M	WH	--	A	PROBAND	688	1	Low serum alkaline phosphatase; high urinary phosphoethanolamine; a sib died at age 5 mos. from same disease; shows bowing of the long bones and abnormal metaphyses; deficient fibroblast alkaline phosphatase activity
GM4234	6	EEMEM 20U	0321	27	F	WH	+-(0)		MOTHER	688	2	Clinically normal; low serum alkaline phosphatase level; two affected children
GM4236	6	EEMEM 20U	0321	38	M	WH	+-(0)		FATHER	688	3	Low serum alkaline phosphatase level; two affected children; clinically normal
GM4263	7	EEMEM 20U	0371	30	F	WH	+-(0)		PARENT	947	1	Affected child first showed symptoms at 3 mos. of age and died at age 1 year; proband not in Repository; 46,XX
Ichthyosis Congenita, Harlequin Fetus Type - 24250												
GM7065	2	EEMEM 20U	0355	1DA	M	WH	--		PROBAND			Thick, cracked yellowish skin broken into plates; swollen hands & feet with hypoplastic digits & very small nails; bilateral ectropion of eyelids; flattened nose & auricles; sparse hair; negative family history
Klippel-Feil Syndrome - 14890												
GM3884	7	EEMEM 10I	0354	14	F	WH	+-	A	PROBAND			Congenital heart murmur; bilaterally enlarged kidneys; mild hearing loss; inguinal hernias; 46,X,+t(X;16)(Xpter>Xq26::16q24>16qter;16pter>16q24::Xq26>Xqter), balanced; normal X is late replicating; slow growing culture
Leprechaunism - 24620												
*GM5241	6	EEMEM 20U	0407	1MO	F	WH	--		PROBAND			Low birth weight; hirsutism, insulin resistant, characteristic facies, & no subcutaneous fat; normal number of insulin receptors with reduced affinity in fibroblasts; reduced number of receptors with normal affinity in lymphoblasts

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Marfan Syndrome - 15470												
*GM0036	4	EEMEM 20U	0026	10	M	WH	+-		PROBAND	112	1	46,XY; kyphoscoliosis; tall; long arms; similarly affected sib
*GM0035	4	EEMEM 20U	0026	12	M	WH	+-		AFF BRO	112	2	46,XY; kyphoscoliosis; tall; long arms
GM7726	2	EEMEM 20U	0107	12	F	BL	+-		PROBAND	1070	1	Similarly affected mother; long face; high arched palate; genu valgum; mild scoliosis; tall; wide arm span; decreased U/L segment ratio; no chest or eye abnormalities; mitral valve prolapse; arachnodactyly; see GM7727A Lymphoid
Metaphyseal Dysplasia, Anetoderma & Optic Atrophy - 25045												
GM3129	3	EEMEM 20U	0232	27	F	WH	--		PROBAND	452	1	Uninvolved skin; macular anetoderma of Schweninger and Buzzi; Jewish; same patient as GM3130
GM3130	2	EEMEM 20U	0232	27	F	WH	--		PROBAND	452	1	Involved skin; same patient as GM3129; macular anetoderma of Schweninger and Buzzi; Jewish
Metatropic Dwarfism, Type II (Kneist Disease) - 15655												
GM4179	2	EEMEM 20U	0006	9MO	F	BL	+-		PROBAND			Short-limbed dwarfism with tracheal papillomas; parents and 4 sibs are clinically normal, one sib died as a premature infant
GM4911	9	EEMEM 20U	0270	20	M	WH	+-		PROBAND			Midface hypoplasia; protuberant eyes; cleft palate; sec. conductive deafness; retinal detachment; myopia; norm. IQ; neg. fam. history, urine MPS excretion norm. but keratan sulfate is elevated & 65% is poorly degraded dermatan sulfate
Winchester Disease; Mucopolysaccharidosis - 27795												
*GM2295	2	EEMEM 20U	0180	21	F	WH	--	B	PROBAND	477	1	Puerto Rican; rheumatoid arthritis; coarse facies; corneal opacity; increased intracellular uronic acid level in fibroblasts; similarly affected sib
Limb-Girdle Muscular Dystrophy (Pelvofemoral, Leyden-Moebius M. D.) - 25360												
GM3932	4	EEMEM 20U	0348	67	M	WH	--		PROBAND	795	1	Myopathic EMG; elevated CPK; onset at age 18; slowly progressive proximal muscle weakness; impotence; deceased male sib had same disorder; 46,XY; see GM3931A Lymphoid
Muscular Dystrophy; Becker Type (Progressive Tardive) - 31010												
GM2298	R	EEMEM 15U	0202	18	M		Y-		PROBAND			Clinically affected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
				DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE								
				Muscular Dystrophy; Becker Type (Progressive Tardive) - 31010								
GM4981	5	EEMEM 15U	0347	6	M	WH	Y-		PROBAND			Significant lordosis; pseudohypertrophy of calves; elevated CPK; two affected maternal uncles
				Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020								
*GM1695	5	EEMEM 20U	0176	26	F	WH	+-	A	PROBAND			46,X,t(X;11)(Xqter>Xp2105::11q13>11qter; 11pter>11q13::Xp2105>Xpter); balanced; in blood leukocytes, the normal X shows non-random inactivation; clinically affected
*GM2339	8	EEMEM 20U	0088	20FW	M		Y?		PROBAND			Presumptive affected fetus (50% risk); 46,XY
*GM3429	10	EEMEM 20U	0049	6	M	WH	Y-		PROBAND			Canadian Repository #466; progressive proximal muscle weakness; elevated CPK; myopathic EMG; negative family history
GM3604A	6	EEMEM 20U	0006	4	M	WH	Y-		PROBAND			Abnormal EMG; abnormal muscle biopsy; elevated CPK; no family history of muscular dystrophies
GM4315	3	EEMEM 20U	0006	41	F	BL	+-(D)		PARENT			Three affected brothers; one affected son; also heterozygous for G6PD (B/A-); elevated serum CPK; proband not in Repository
*GM5835	6	EEMEM 20U	0426	10	F	OT	+-	A	PROBAND			Chinese-Hawaiian-Caucasian; clinically norm, parents & 3 male sibs; short stature; elev. CPK; muscle atrophy & weakness; de novo trans.; 46,X,t(X;5)(Xqter> Xp21.2::5q35.3>5qter;5pter>5q35.3:: Xp21.2>Xpter); bal.; norm X is late repl
GM3781	3	EEMEM 20U	0006	11	M	WH	Y-		PROBAND	533	1	Latin American; elevated serum CPK; similarly affected sib; see GM3780 Lymphoid
*GM3783	2	EEMEM 20U	0006	10	M	WH	Y-		AFF BRO	533	2	Latin American; elevated serum CPK; see GM3782 Lymphoid
GM4327	3	EEMEM 20U	0125	23	M	WH	Y-		PROBAND	708	2	Sardinian; normal G6PD; one affected brother and one affected sororal nephew
GM5104	2	EEMEM 20U	0125		F	WH	+-(D)		SIS	708	3	Sardinian; double heterozygote for DMD & G6PD deficiency of the Mediterranean type; one affected son (G6PD norm), and one unaffected son (G6PD deficient), and two affected brothers (G6PD norm)

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE							
Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020											
*GM4978A	7	EEMEM 20I	0347	13	M	BL	Y-	PROBAND	805	1	Affected brother and male cousin; muscle biopsy shows advanced dystrophic process; elevated serum CPK; pseudohypertrophy of calves; proximal muscle weakness
GM4982	4	EEMEM 15U	0347	34	F	BL	+-(O)	MOTHER	805	2	Clinically unaffected; normal CPK; two affected sons & one affected nephew
GM5162	2	EEMEM 20U	0348	13	M	WH	Y-	PROBAND	824	1	Progressive weakness; pseudohypertrophy; similarly affected brother & male maternal cousins; 46,XY; see GM3929A Lymphoid
GM5158	3	EEMEM 20U	0348	47	F	WH	+-(O)	MOTHER	824	2	Clinically unaffected; 2 affected sons & 2 affected great nephews; see GM5159 Lymphoid
GM5160	2	EEMEM 20U	0348	57	M	WH	++	FATHER	824	3	Clinically unaffected; 2 affected sons; see GM5161A Lymphoid
Myositis Ossificans Progressiva - 13510											
GM0513	3	EEMEM 20U	0008	16	M	WH	+-	PROBAND			Clinically affected
GM0783	3	EEMEM 20U	0107		M	BL	+-	PROBAND			Clinically affected
Myotonic Dystrophy (Steinert Disease) - 16090											
GM3132	2	EEMEM 20U	0210	18	M	WH	+-	PROBAND			Bone deformities; muscle wasting and myotonia
*GM3755	3	EEMEM 20U	0348	44	M	WH	+-	PROBAND	488	1	Typical phenotype; mother, two brothers and a sister also affected; see GM3756 Lymphoid
GM3987	2	EEMEM 20U	0348	39	M	WH	+-	AFF BRO	488	2	Distal weakness; clinical and electrical myotonia; cataracts; marked balding; see GM3986A Lymphoid
GM3989	2	EEMEM 20U	0348	38	M	WH	+-	AFF BRO	488	3	Distal weakness; frontal baldness; cataracts; clinical and electrical myotonia
GM3991	3	EEMEM 20U	0348	76	F	WH	+-	AFF MOTHER	488	4	Distal weakness; cataracts; clinical and electrical myotonia; frontal baldness; see GM3990A Lymphoid
GM3759	4	EEMEM 20U	0348	33	M	WH	+-	PROBAND	527	1	EMG myotonia; myotonic face; distal weakness and atrophy; positive family history; affected daughter is GM3696A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE							
Myotonic Dystrophy (Steinert Disease) - 16090											
GM4033	2	EEMEM 20U	0348	48	M	WH	+-	AFF FATHER	590	2	Temporal atrophy; clinical myotonia; distal weakness; positive family history; see GM4034 Lymphoid; proband is GM3697A Lymphoid
Myopathy, Unusual Family With Central Nuclei, Recessive or X-linked Inher.											
GM6813	2	EEMEM 20U	0462	1MO	M	WH		PROBAND			Hypotonia; respiratory distress; low birth weight; cryptorchidism; large simple ears; mild contractures of knees; overlapping fingers; micrognathia; 46,XY in lymphocytes and fibroblasts; similarly affected brother
Myotonic Chondrodystrophy - Schwartz-Jampel-Aberfeld Syndrome - 25580											
GM4465	2	EEMEM 20U	0204	7	F	WH	--	PROBAND			Short stature; joint limitations; myotonia; muscle hypertrophy; typical facial features; only affected family member
Osteopetrosis with Renal Tubular Acidosis - 25973											
*GM7270	2	EEMEM 20U	0319	19	M	WH	--	PROBAND			Deficient RBC carbonic anhydrase II activity; parents are third cousins; short stature; anemia; hepatosplenomegaly; proximal and distal tubular acidosis; generalized osteopetrosis
*GM6071	5	EEMEM 20U	0126	25	F	WH	--	AFF SIS	904	2	Diagnosed at age 1; short stature, multiple fractures, dull mentality, dental malocclusion, bilateral optic atrophy, high arched palate, and mild hirsutism; deficient RBC carbonic anhydrase II activity; see GM6065 Lymphoid
*GM6072	6	EEMEM 20U	0126	54	F	WH	+-(O)	MOTHER	904	3	Three affected daughters and one unaffected daughter; asymptomatic; 50% of normal RBC carbonic anhydrase II activity; slow growing culture; see GM6067 Lymphoid; proband is GM6066 Lymphoid
*GM6073	3	EEMEM 20U	0126	56	M	WH	+-(O)	FATHER	904	4	Three affected daughters and one unaffected daughter; asymptomatic; 50% of normal RBC carbonic anhydrase II activity; see GM6068 Lymphoid; proband is GM6066 Lymphoid
Osteogenesis Imperfecta: Autosomal Dominant - 16621; 16620; 16622; 16623											
GM5181	6	EEMEM 20U	0404	25	M	WH	+-	PROBAND	826	1	Type I; blue sclerae, fractures, & normal hearing; 3 sibs & mother are affected; French-Canadian

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Osteogenesis Imperfecta: Autosomal Dominant - 16621, 16620, 16622, 16623												
GM5182	5	EEMEM 20U	0404	46	F	WH	+-		AFF MOTHER	826	2	Type I; blue sclerae, fractures, & hearing loss; 4 affected children; French-Canadian
Osteogenesis Imperfecta: Autosomal Recessive - 25940, 25941, 25942												
GM4178	2	EEMEM 20U	0122	2MO	F	WH	--		PROBAND			Parents clinically normal; neonatal lethal form; blue sclera; fractures of extremities; normal bone shape; similarly affected sib
Osteogenesis Imperfecta: Genetics Uncertain; Non-Lethal												
GM1093	7	EEMEM 20U	0095	8	M	WH	-?		PROBAND			Normal fibroblast alkaline phosphatase activity; clinically affected
GM1436A	6	EEMEM 20U	0147	24	M	WH	-?		PROBAND			Normal fibroblast alkaline phosphatase activity; clinically affected
GM1789	2	EEMEM 20U	0147	12	F	WH	-?		PROBAND			Scoliosis; slightly blue sclerae; negative family history; normal fibroblast alkaline phosphatase activity
GM2573	3	EEMEM 20U	0147	17	M	WH	-?		PROBAND			Normal fibroblast alkaline phosphatase activity; clinically affected
GM2645	2	EEMEM 20U	0147	17	M	WH	-?		PROBAND			Positive family history; normal fibroblast alkaline phosphatase activity; clinically affected
GM2695	2	EEMEM 20U	0147	12	F	WH	-?		PROBAND			Normal fibroblast alkaline phosphatase activity; clinically affected
GM3138	2	EEMEM 20U	0147	3	F	WH	-?		PROBAND			Broken bones; opalescent teeth; blue sclera; poor motor development; normal fibroblast alkaline phosphatase activity
Osteogenesis Imperfecta: Genetics Uncertain; Lethal												
*GM0744	6	EEMEM 20U	0057	1DA	F	WH	-?		PROBAND			46,XX; negative family hx; multiple fractures of extremities, skull, & ribs present at birth; lethal; normal fibroblast alkaline phosphatase activity
*GM2328	2	EEMEM 20U	0107	4DA	M	WH	-?		PROBAND			First affected child of clinically normal parents; normal fibroblast alkaline phosphatase activity
*GM2962	3	EEMEM 20U	0070	8DA	F	WH	-?		PROBAND			"Broad boned"; normal fibroblast alkaline phosphatase activity; neonatal lethal form

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Osteogenesis Imperfecta: Genetics Uncertain; Lethal												
GM5747A	7	EEMEM 20U	0078	6WK	M	WH	-?		PROBAND			Colombian; blue sclerae; pliable skull; poor mineralization of skull and short, thick, ribbon-like poorly mineralized long bones; multiple fractures; normal serum phosphoethanolamine
GM6260	9	EEMEM 20U	00	5MO	M	WH	-?		PROBAND			Negative family history; short arms and legs; large head; multiple fractures of ribs and extremities present at birth; telangiectasias on cheeks; soft skull bones; blue sclerae bilaterally
Parana Hard-Skin Syndrome - 26053												
GM2663	2	EEMEM 20U	0147	13	M		--		PROBAND			Clinically affected
Pseudoachondroplastic Dysplasia IV - 26416												
GM3127	2	EEMEM 20U	0107	3	F		--		PROBAND			Sibling died of same disorder
SC Phocomelia - Roberts Syndrome: 26900 - 26830												
*GM3913	12	EEMEM 20I	0356	20MO	M	WH	--	A	PROBAND			French Canadian; absent radii and short ulnae; microphthalmia; corneal opacity; chromatid breaks and gaps; centromere splitting & puffing; 46% 46,XY/ 54% 47, XY,+7; 48% tetraploid at P15
Schinzel-Giedion Syndrome - 26915												
*GM6172	7	EEMEM 20U	0437	4MO	F	WH	--		PROBAND			Negative family history; coarse facies, frontal bossing, midface retraction, hypertelorism, facial hemangiomata, short up-turned nose, and unusually malformed ears; generalized skeletal dysplasia; bilateral hydronephrosis
Thanatophoric Dwarfism - 18760												
GM1422	2	EEMEM 20U	0107	2WK	F	BL	+-		PROBAND			Severely dwarfed; extremely short extremities; small hands and feet; large head; distended abdomen; normal fibroblast alkaline phosphatase activity; negative family history
GM0711	2	EEMEM 20U	0143	1DA	M	WH	+-		PROBAND	182	1	Fibroblast culture; cartilage; clinically affected
GM0712	2	EEMEM 20U	0143	1DA	M	WH	+-		PROBAND	182	1	Fibroblast culture; spleen; same pt. as GM0711
GM0713	2	EEMEM 20U	0143	1DA	M	WH	+-		PROBAND	182	1	Fibroblast culture; lung; same pt. as GM0711

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Thanatophoric Dwarfism - 18760												
GM0714	3	EEMEM 20U	0143	10A	M	WH	+-		PROBAND	182	1	Fibroblast culture; skin; same pt. as GM0711
Werdnig-Hoffmann Disease - 25330												
*GM0232	2	EEMEM 20U	0078	7MO	M	WH	--		PROBAND			Progressive muscular atrophy; abnormal EMG; absent deep tendon reflexes
GM3815	2	EEMEM 20U	0348		M	WH	+-(O)		FATHER	553	1	Clinically normal; two affected children
GM3814	2	EEMEM 20U	0348		F	WH	+-(O)		MOTHER	553	2	Clinically normal; two affected children
GM3813	2	EEMEM 20U	0348	3	M	WH	--		PROBAND	553	3	Marked muscle atrophy and weakness; similarly affected brother
DISORDERS OF THE NERVOUS SYSTEM												
Apparently Normal Individuals With Normal Psychiatric Evaluations												
*GM4501A	8	EEMEM 20U	0191	19	M	WH			PROBAND	730	1	Line HF20; monozygotic twin of GM4502A; 46,XY
*GM4502A	11	EEMEM 20U	0191	19	M	WH			IDENT TWIN	730	2	Line HF21; monozygotic twin of GM4501A; 46,XY; 10% of cells show random chromosome loss
*GM4503B	9	EEMEM 20U	0191	31	F	WH			PROBAND	731	1	Line HF22; monozygotic twin of GM4504A; has had some minor depression; 46,XX
*GM4504A	7	EEMEM 20U	0191	31	F	WH			IDENT TWIN	731	2	Line HF23; monozygotic twin of GM4503B; 46,XX; 13% of cells show random chromosome loss
*GM4505A	6	EEMEM 20U	0191	20	F	WH			PROBAND	732	1	Line HF24; monozygotic twin of GM4506A; 46,XX; 6% of cells show random chromosome loss
*GM4506A	7	EEMEM 20U	0191	20	F	WH			IDENT TWIN	732	2	Line HF25; monozygotic twin of GM4505A; 46,XX/46,XX,t(13;14)(13pter>13q34::14q21>14qter::14pter>14q21::13q34>13qter); 66%/30%; 4% of cells show random chromosome loss
GM7492	1	EEMEM 20U	0308	17	M	WH			PROBAND	1049	1	See GM7491 Lymphoid; 46,XY; 16% of cells show random chromosome loss
GM7522	2	EEMEM 20U	0308	19	F	WH			PROBAND	1051	1	See GM7521 Lymphoid; 46,XX; 12% of cells show random chromosome loss
GM7545	5	EEMEM 20U	0308	22	F	WH			PROBAND	1055	1	See GM7544 Lymphoid; 46,XX; 10% of cells show random chromosome loss

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
			Apparently Normal Individuals With Normal Psychiatric Evaluations								
GM7532	3	EEMEM 20U	0308	16	F	OT		PROBAND	1057	1	Caucasian mother and Japanese father; see GM7533 Lymphoid; 46,XX; 8% of cells show random chromosome loss
GM7525	3	EEMEM 20U	0308	22	F	WH		PROBAND	1059	1	See GM7524 Lymphoid; 46,XX; 10% of cells show random chromosome loss
GM7753	2	EEMEM 20U	0308	17	M	WH		PROBAND	1071	1	See GM7752 Lymphoid; 46,XY; 6% of cells show random chromosome loss
			Alzheimer Disease of Brain - 10430								
*GM03648	6	EEMEM 20U	0139	53	M	WH	+-	PROBAND	87	1	Pedigree contains many affected individuals; 46,XY; clinically affected
GM0490	3	EEMEM 20U	0139	33	F	WH	+?	NIECE	87	6	At risk
			Azorean Neurologic Disease: Machado-Joseph Disease - 10915								
GM6151	3	EEMEM 20U	0422	38	M	WH	+-	PROBAND	936	1	Portuguese; affected father and 2 sibs; Type III: ataxia, nystagmus, and dysarthria
GM6153	2	EEMEM 20U	0422	44	M	WH	+-	AFF BRO	936	2	Portuguese; affected father and 2 sibs; Type III: slight ataxia, dysarthria, and nystagmus
			Canavan Disease (Spongy Degeneration of the CNS) - 27190								
GM0059	2	EEMEM 20U	0095	17MO	F		--	PROBAND	424	1	Similarly affected sib
GM0060	4	EEMEM 20U	0095	2	M		--	AFF BRO	424	2	Brain tissue fibroblast culture
			Charcot-Marie Tooth Disease (Peroneal Muscular Atrophy, Axonal Type) - 11820								
*GM4598	5	EEMEM 20U	0260	30	M	WH	+-	PROBAND	746	1	Hypertrophic variety of C-M-T disease; pedigree shows dominant inheritance; diagnosis confirmed by nerve conductivity tests; 46,XY,9qh+/47,XY,+7,9qh+; 80%/20%
*GM4597	5	EEMEM 20U	0260	56	F	WH	+-	AFF MOTHER	746	2	Hypertrophic variety of C-M-T disease; pedigree shows dominant inheritance; diagnosis confirmed by nerve conductivity tests
*GM4594	6	EEMEM 20U	0260	36	F	WH	+-	AFF SIS	746	3	Mother of GM4595; hypertrophic variety of C-M-T disease; pedigree shows dominant inheritance; diagnosis confirmed by nerve conductivity tests
GM4593	5	EEMEM 20U	0260	37	M	WH	++	BRO-IN-LAW	746	4	Clinically normal father of GM4595; spouse of GM4594

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
Charcot-Marie Tooth Disease (Peroneal Muscular Atrophy, Axonal Type) - 11820											
*GM4595	5	EEMEM 20U	0260	9	F	WH	+-	AFF NIECE	746	5	Hypertrophic variety of C-M-T disease; pedigree shows dominant inheritance; diagnosis confirmed by nerve conductivity tests
*GM4596	5	EEMEM 20U	0260	83	F	WH	+-	AFF GR AUNT	746	6	Hypertrophic variety of C-M-T disease; pedigree shows dominant inheritance; diagnosis confirmed by nerve conductivity tests
Dysautonomia (Riley-Day Syndrome) - 22390											
GM0732	2	EEMEM 20U	0107	15MO	M	WH	--	PROBAND			Non-Jewish; more severe than classical form
*GM0850A	8	R1640 20U	0025	26	M	WH	--	PROBAND			Clinically affected
*GM2341	7	EEMEM 10U	0191	17	M		--	PROBAND	98	1	Mild familial type; similarly affected sib
*GM2342	6	EEMEM 10U	0191	19	M		--	AFF BRO	98	2	Mild familial type
GM4623	3	EEMEM 20U	0074	38	F	WH	+-(O)	PARENT	759	2	Jewish; mother of an affected child; proband not in Repository
GM4624	3	EEMEM 20U	0074	39	M	AI	+-(O)	PARENT	759	3	Father of an affected child; proband not in Repository
*GM2343	6	EEMEM 10U	0191	24	F	WH	--	PROBAND	761	1	Mild familial type
GM4632	3	EEMEM 20U	0110	58	M	WH	+-(O)	FATHER	761	2	Clinically normal; Jewish (Ashkenazi)
GM4634	2	EEMEM 20U	0110	53	F	WH	+-(O)	MOTHER	761	3	Clinically normal; Jewish (Ashkenazi); see GM4635 Lymphoid
GM4638	12	EEMEM 20U	0191	7	F	WH	--	PROBAND	762	1	Line HF63; Ashkenazi; alacrima, recurrent pneumonias, emotional immaturity, chronic dehydration, abotemia, anemia, poor muscle development, & absent DTR's
GM4637	13	EEMEM 20U	0191	32	F	WH	+-(O)	MOTHER	762	2	Clinically normal; line HF62; Ashkenazi
GM4663	5	EEMEM 20U	0191	2	F	WH	--	PROBAND	763	1	Line HF65; Ashkenazi; smooth tongue, absent DTR's, bilateral corneal scars, sluggish corneal reflexes, developmental delay, negative histamine test, & erythematous blotching of the skin
GM4664	9	EEMEM 20U	0191	32	M	WH	+-(O)	FATHER	763	2	Line HF66; Ashkenazi; clinically unaffected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Dysautonomia (Riley-Day Syndrome) - 22390											
GM4665	8	EEMEM 20U	0191	30	F	WH	+-(O)	MOTHER	763	3	Line HF67; Ashkenazi; clinically unaffected
Fahr's Disease: Cerebral Calcification, Non-Arteriosclerotic - 21360											
*GM3242	2	EEMEM 20U	0078	3	F		--	PROBAND			46,XX; a sister is also affected; periventricular calcifications; abnormal EEG; developmental delay; glaucoma
Friedreich Ataxia - 22930 + 13660											
*GM1960	5	EEMEM 20U	0105	15	F	WH		PROBAND			Mexican; marked weakness, hypotonia, cerebellar deficit, and sensory deficit; abolished muscle stretch reflexes; similarly affected male sib; slow growing culture
GM3665		EEMEM 15U	0344	13	F	WH		PROBAND			Limb and gait ataxia; scoliosis; proprioceptive sensory loss; 46,XX; negative family history; recessive inheritance; passage 3 at I4R
*GM3295	3	EEMEM 20U	0210	63	M	WH		PROBAND	68	1	Son and father also affected; pes cavus; peripheral neuropathy; cerebellar ataxia; nystagmus; sensorineural hearing loss; possibly dominant inheritance; see GM3959 Lymphoid
GM3816	2	EEMEM 20U	0348	36	F	WH		PROBAND	594	1	Spinal-cerebral degeneration with myocardiopathy; a brother is similarly affected; probably recessive inheritance
GM4078	2	EEMEM 20U	0348	30	M	WH		AFF BRO	594	2	Ataxia, cardiomyopathy, and mild peripheral neuropathy; see GM4079 Lymphoid; probably recessive inheritance
Gilles de la Tourette Syndrome - 13750											
GM2642	11	EEMEM 10U	0191	12	M		+-	PROBAND			No family history of disease; progressive movement disorder since age 4; symptoms include extraordinary & violent actions
GM2664	10	EEMEM 20U	0191	11	M		+-	PROBAND	523	1	Positive family history; abnormal body movements since age 3; formerly GM2643
Huntington's Disease - 14310											
*GM1168	3	EEMEM 20U	0136	43	M	WH	+-	PROBAND			Slurred speech; upper extremity chorea; no family history available
*GM1169	2	EEMEM 20U	0136	50	M	WH	+-	PROBAND			Choreoathetotic movements of all extremities and slurred speech; positive family history of H.D.

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease - 14310												
*GM1187	3	EEMEM 20U	0136	43	M	WH	+-		PROBAND			Slurred speech; left upper extremity dysmetria; unsteady gait; positive family history of H.D.
*GM1171	3	EEMEM 20U	0136	37	F	WH	+?		PROBAND	106	1	At risk (50%); a sister has H.D.
*GM2079	3	EEMEM 20U	0136	48	F	WH	+-		PROBAND	107	1	See GM2080A Lymphoid; father is also affected
*GM2077	7	EEMEM 20U	0136	25	F	WH	+?		DAU	107	2	At risk (50%); mother and grandfather are affected; see GM2078 Lymphoid
GM2147A	11	EEMEM 20U	0186	55	M	WH	+-		PROBAND	108	1	Mother was also affected; see GM2146 Lymphoid
*GM2149	3	EEMEM 20U	0186	54	F	WH	++		SPOUSE	108	2	Normal; see GM2148 Lymphoid
*GM2151	3	EEMEM 20U	0186	26	F	WH	+?		DAU	108	3	At risk (50%); see GM2150 Lymphoid; personality disorder but no chorea
GM6274	2	EEMEM 20U	0186	56	F	WH	+-		AFF MAT COU	108	4	Chorea; depression; loss of temper; positive family history; sister of GM6273
GM6273	2	EEMEM 20U	0186	51	F	WH	+?		MAT COU	108	5	Clinically unaffected; sister of GM6274; reduced at risk
GM2153	3	EEMEM 20U	0186	40	F	WH	++		SPOUSE	109	2	Clinically normal widow of affected husband; proband not in Repository; see GM2152A Lymphoid; 46,XX; 10% of cells show random chromosome loss
GM2155	3	EEMEM 20U	0186	20	F	WH	+?		DAU	109	3	At risk (50%); father had H.D.; see GM2154B Lymphoid
GM2157	4	EEMEM 20U	0186	16	F	WH	+?		DAU	109	4	At risk (50%); father had H.D.; see GM2156 Lymphoid
GM2159	3	EEMEM 20U	0186	19	F	WH	+?		DAU	109	5	At risk (50%); father had H.D.; see GM2158 Lymphoid
GM2161	3	EEMEM 20U	0186	21	M	WH	+?		SON	109	6	At risk (50%); father had H.D.; see GM2160 Lymphoid
GM2163	3	EEMEM 20U	0186	12	M	WH	+?		SON	109	7	At risk (50%); father had H.D.; see GM2162 Lymphoid
*GM2165	2	EEMEM 20U	0186	55	M	WH	+-		PROBAND	110	1	Mother and sister were also affected; age of onset 46 years; see GM2164B Lymphoid; 46,XY

BIOCHEMICALLY MUTANT ~~HUMAN~~ FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease - 14310											
*GM2177	6	EEMEM 20U	0186	26	M	WH	+?	SON	110	2	At risk (50%); see GM2176 Lymphoid; may show early chorea; has long standing personality problem
*GM2183	3	EEMEM 20U	0186	21	F	WH	+?	DAU	110	3	At risk (50%); see GM2182 Lymphoid
*GM2169	3	EEMEM 20U	0186	52	M	WH	++	BRO-IN-LAW	110	4	Normal spouse of GM2167A; see also GM2168A Lymphoid; 46,XY; 10% of cells show random loss of 1 chromosome
GM2187	3	EEMEM 20U	0186	60	F	WH	+?	SIS	110	5	Asymptomatic at age 60; reduced at risk; see GM2186 Lymphoid
GM2189	2	EEMEM 20U	0186	63	M	WH	++	BRO-IN-LAW	110	6	Normal spouse of GM2187; see GM2188 Lymphoid; 46,XY; 10% of cells show random chromosome loss
*GM2167A	5	EEMEM 20U	0186	61	F	WH	+-	AFF SIS	110	7	See GM2166B Lymphoid; mother and brother are also affected
*GM3621	4	EEMEM 20U	0186	29	F	WH	+-	AFF DAU	110	10	See GM3620 Lymphoid; age of onset 18 years; similarly affected father and grandmother; 46,XX
*GM2173A	8	EEMEM 20U	0186	52	F	WH	+-	PROBAND	111	1	46,XX; see GM2172A Lymphoid; clinically affected
*GM2171	2	EEMEM 20U	0186	22	F	WH	+?	DAU	111	2	At risk (50%); see GM2170 Lymphoid
*GM2175	2	EEMEM 20U	0186	55	M	WH	++	SPOUSE	111	3	See GM2174 Lymphoid; clinically normal; 46,XY; 12% of cells show random chromosome loss/gain; 10% of cells have t(1;1)
GM2123	2	EEMEM 20U	0190	7FW	M	WH	+?	PROBAND	206	1	Fetus at 25% risk for Huntington's disease; 46,XY
GM2191	3	EEMEM 20U	0190	32	F	WH	+?	MOTHER	206	2	At risk (50%); see GM2190 Lymphoid
GM1136A	10	EEMEM 20U	0136	45	M	WH	+-	PROBAND	286	1	Clinically affected
*GM0305A	9	EEMEM 20U	0305	56	F	WH	+-	PROBAND	288	1	Onset at age 46; uncertain family history; formerly GM0094; normal spouse is GM0288A Fibroblast
*GM1061A	5	EEMEM 20U	0136	51	M	WH	+-	PROBAND	292	1	Choreoathetosis, dysarthria, & dysphagia; positive family history; see GM3642 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease - 14310												
*GM1065	3	EEMEM 20U	0136	44	M	WH	+-		PROBAND	293	1	Affected mother & grandmother; moderate ataxia, chorea, dysarthria, and dysphagia; see GM3644 Lymphoid; onset at age 35
GM1083A	8	EEMEM 20U	0136	47	F	WH	+-		AFF SIS	293	2	See GM3643 Lymphoid; choreoathetosis, dysarthria, and dysphagia; onset at age 35 yrs
GM3791	7	EEMEM 20U	0186	15	M	WH	+?		SON	549	2	At risk (50%); proband is GM3790 Lymphoid
GM3793	6	EEMEM 20U	0186	19	F	WH	+?		DAU	549	3	At risk (50%); see GM3794 Lymphoid
GM3860	4	EEMEM 20U	0320	58	F	WH	+?		SIS	568	1	Reduced risk for H.D.; had ovarian cancer at age 50; see GM3861 Lymphoid
GM3870	7	EEMEM 20U	0320	56	F	WH	+?		SIS	568	2	Reduced at risk; also had malignant melanoma; see GM3871 Lymphoid
GM3864A	4	EEMEM 20U	0320	51	F	WH	+-		PROBAND	568	3	Similarly affected sib; see GM3865 Lymphoid
GM3872	5	EEMEM 20U	0320	31	M	WH	+?		SON	568	4	At risk (50%); mother had H.D.; see GM3873 Lymphoid
GM3866	3	EEMEM 20U	0320	26	F	WH	+?		DAU	568	5	At risk (50%); mother had H.D.; see GM3867 Lymphoid
GM3868	5	EEMEM 20U	0320	45	F	WH	+-		AFF SIS	568	6	See GM3869 Lymphoid; clinically affected
GM3862	3	EEMEM 20U	0320	24	F	WH	+?		NIECE	568	7	At risk (50%); daughter of GM3868; see GM3863 Lymphoid
GM4022	3	EEMEM 20U	0136	28	F	WH	+?		DAU	587	4	At risk (50%); mother had H.D.; see GM4023 Lymphoid; proband is GM4017 Lymphoid
*GM4188	3	EEMEM 20U	0367	77	F	WH	++		SPOUSE	682	2	Clinically normal; see GM4189 Lymphoid; proband is GM4219 Lymphoid; 46,XX/46,XX, t(6;19)(6pter>6q13::19p13>19pter;19qter >19p13::6q13>6qter); 78%/22%
*GM4196	2	EEMEM 20U	0367	51	F	WH	+-		AFF DAU	682	3	See GM4197 Lymphoid; clinically affected
*GM4200	2	EEMEM 20U	0367	53	M	WH	+-		AFF SON	682	5	See GM4201A Lymphoid; clinically affected

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease - 14310											
*GM4190	2	EEMEM 20U	0367	85	F	WH	++	SIS-IN-LAW	682	6	Clinically normal; see GM4191 Lymphoid; mother of GM4194, 4198, and 4222; 46,XX, 1qh+
*GM4198	2	EEMEM 20U	0367	63	F	WH	+-	AFF NIECE	682	7	Daughter of GM4190; see GM4199 Lymphoid; father had H.D.
*GM4194	3	EEMEM 20U	0367	60	F	WH	+-	AFF NIECE	682	8	Daughter of GM4190; see GM4195 Lymphoid; father had H.D.
*GM4222	3	EEMEM 20U	0367	59	M	WH	+-	AFF NEPH	682	9	Son of GM4190; see GM4223 Lymphoid; father had H.D.
*GM4226	5	EEMEM 20U	0367	74	M	WH	+?	BRO	682	10	Reduced at risk; see GM4227 Lymphoid; mother had H.D.
*GM4232	3	EEMEM 20U	0367	70	M	WH	+?	BRO	682	11	Reduced at risk; see GM4233 Lymphoid; mother had H.D.
*GM4192	4	EEMEM 20U	0367	71	M	WH	+?	1ST COU	682	13	Reduced at risk; mother had H.D.; see GM4193 Lymphoid
*GM4206	4	EEMEM 20U	0367	75	F	WH	+?	1ST COU	682	14	Reduced at risk; mother had H.D.; see GM4207 Lymphoid
*GM4204	4	EEMEM 20U	0367	81	M	WH	++	COU-IN-LAW	682	15	Related by marriage; clinically normal; father of GM4210, 4212, and 4230; see GM4205 Lymphoid; 46,XY
*GM4212	2	EEMEM 20U	0367	50	F	WH	+-	AFF COU	682	19	Early symptoms of H.D.; daughter of GM4204; see GM4213 Lymphoid; mother had H.D.; 1st cousin once removed to proband
*GM4230	2	EEMEM 20U	0367	55	M	WH	+-	AFF COU	682	20	Mother had H.D.; daughter is GM4220; father is GM4204; see GM4231 Lymphoid; 1st cousin once removed to proband
*GM4210	3	EEMEM 20U	0367	58	M	WH	+-	AFF COU	682	21	Father of GM4208; see GM4211 Lymphoid; mother had H.D.; 1st cousin once removed to proband
*GM4208	2	EEMEM 20U	0367	35	M	WH	+-	AFF COU	682	22	Son of GM4210; see GM4209 Lymphoid; 1st cousin twice removed to proband
*GM4220	3	EEMEM 20U	0367	31	F	WH	+?	COU	682	23	At risk (50%); daughter of GM4230; see GM4221 Lymphoid; 1st cousin twice removed to proband; father had H.D.

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease - 14310											
*GM5031B	2	EEMEM 20U	0367	60	M	WH	+-	AFF COU	682	26	Choreic movements & dementia; first cousin once removed to proband; see GM4894 Lymphoid
*GM5030B	3	EEMEM 20U	0367	56	M	WH	+-	AFF COU	682	27	Choreic movements & dementia; first cousin once removed to proband; see GM4915A Lymphoid
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4281	2	EEMEM 20U	0233	21	F	WH	+-?	PROBAND	690	1	Code 48; rigid form of HD; possible homozygote; both parents affected; age of onset 14 yrs; neuro exam 3/82 shows a hypokinetic variant of HD with dystonia and marked tremor, ataxic wide-based gait; see GM4282 Lymphoid
*GM4867	4	EEMEM 20U	0233	49	M	WH	+-	AFF FATHER	690	2	Code 47; clinically affected; neuro exam 3/81 shows rigidity and a narrow based gait with pauses; see GM4866A Lymphoid
GM5735Z		FRZBX	0233		M	WH	+?	MAT UNCLE	690	3	Code 1173; unaffected; last examined on 4/6/82; father had HD; see GM5736A Lymphoid
GM5725Z		FRZBX	0233	48	F	WH	+?	MAT AUNT	690	4	Code 1165; unaffected; last examined on 4/6/82; father had HD; see GM5726A Lymphoid
GM6684Z		FRZBX	0233		M	WH	+?	MAT UNCLE	690	5	Code 70; unaffected; last examined on 3/29/83; father had HD; affected spouse is GM4767; affected daughter is GM6596Z; see GM6685 Lymphoid
GM4773	2	EEMEM 20U	0233	57	F	WH	+?	MAT AUNT	690	6	Code 441; unaffected; last examined on 3/27/81; two affected parents; spouse of HD patient; affected son is GM4807; see GM4772A Lymphoid
GM4807	2	EEMEM 20U	0233	41	M	WH	+-	AFF 1ST COU	690	7	Code 442; mother is GM4773; father had HD; neuro exam 3/82 shows prominent dyskinesia, fluid eye movements, facial grimacing, and problems swallowing; see GM4284 Lymphoid
GM4719	3	EEMEM 20U	0233	39	F	WH	+?	MAT 1ST COU	690	8	Code 453; unaffected; last examined on 3/23/81; mother is GM4773; father had HD; affected brother is GM4807; see GM4720A Lymphoid

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM4703	5	EEMEM 20U	0233	35	F	WH	+?		MAT 1ST COU	690	9	Code 447; unaffected; last examined on 3/23/81; mother is GM4773; father had HD; affected brother is GM4807; see GM4704A Lymphoid
*GM4805	3	EEMEM 20U	0233	34	F	WH	+?		MAT 1ST COU	690	10	Code 456; unaffected; last examined on 3/27/81; mother is GM4773; father had HD; affected brother is GM4807; see GM4804A Lymphoid
GM5583Z		FRZBX	0233	23	M	WH	+?		MAT 1ST COU	690	11	Code 450; unaffected; last examined on 3/26/82; mother is GM4773; father had HD; affected brother is GM4807; see GM5584A Lymphoid
GM5589Z		FRZBX	0233	44	M	WH	+?		MAT COU	690	12	Code 483; unaffected; last examined on 3/26/82; stepfather and 1st cousin once removed to proband; brother is GM5579Z; unaffected father is GM6682Z; see GM5590A Lymphoid
GM6369Z		FRZBX	0233	30	F	WH	+?		MAT 2ND COU	690	13	Code 386; unaffected; last examined on 3/8/83; father had HD; see GM6370 Lymphoid
GM6782Z		FRZBX	0233	29	F	WH	+?		MAT 2ND COU	690	14	Code 1192; unaffected; last examined on 4/4/83; father had HD
GM5495Z		FRZBX	0233	22	M	WH	+?		MAT 2ND COU	690	15	Code 1195; unaffected; last examined on 3/15/82; father had HD; see GM5496A Lymphoid
GM6768Z		FRZBX	0233	21	F	WH	+?		MAT 2ND COU	690	16	Code 1196; unaffected; last examined on 4/4/83; father had HD; see GM6769 Lymphoid
*GM4777	3	EEMEM 20U	0233	53	M	WH	+-		AFF MAT COU	690	19	Code 608; clinically affected; expired in 1983; affected son is GM4737; see GM4776 Lymphoid
GM4737	3	EEMEM 20U	0233	20	M	WH	+-		AFF 2ND COU	690	21	Code 612; clinically affected; age of onset 18 yrs; affected father is GM4777; neuro exam 3/82 shows choreiform movements; see GM4738A Lymphoid
GM6383Z		FRZBX	0233	30	F	WH	+?		MAT 2ND COU	690	22	Code 613; unaffected; last examined on 3/8/83; affected father is GM4777; affected brother is GM4737; see GM6384 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM5637Z		FRZBX	0233	27	F	WH	+?	MAT 2ND COU	690	23	Code 614; unaffected; last examined on 3/26/82; affected father is GM4777; affected brother is GM4737; see GM5638A Lymphoid
GM5441Z		FRZBX	0233	13	F	WH	+?	MAT 2ND COU	690	24	Code 615; unaffected; last examined on 3/15/82; affected father is GM4777; affected brother is GM4737; see GM5442 Lymphoid
GM5439Z		FRZBX	0233	18	F	WH	+?	MAT 2ND COU	690	25	Code 616; unaffected; last examined on 3/15/82; affected father is GM4777; affected brother is GM4737; see GM5440B Lymphoid
GM5505Z		FRZBX	0233	18	F	WH	+?	MAT 2ND COU	690	26	Code 617; unaffected; last examined on 3/15/82; affected father is GM4777; affected brother is GM4737; see GM5506 Lymphoid
GM6365Z		FRZBX	0233	61	M	WH	+?	MAT 1ST COU	690	27	Code 606; unaffected; last examined on 3/8/83; affected sibs are GM4777 and GM5560B Lymphoid; 1st cousin once removed to proband; see GM6366 Lymphoid
GM6680Z		FRZBX	0233	61	F	WH	+?	MAT COU	690	28	Code 1113; unaffected; last examined on 3/29/83; mother had HD; sibs are GM5691Z and GM6628Z; see GM6681 Lymphoid
GM4707A	3	EEMEM 20U	0233	49	F	WH	+?	MAT COU	690	29	Code 11; unaffected; last examined on 3/23/81; mother is GM5691Z; spouse of GM4711; see GM4708A Lymphoid
*GM4711	2	EEMEM 20U	0233	51	M	WH	+?	MAT COU	690	30	Code 10; unaffected; last examined on 3/23/81; spouse of GM4707A; affected sib is GM4476
GM6343Z		FRZBX	0233	32	M	WH	+?	COU	690	31	Code 1086; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6390Z		FRZBX	0233	31	M	WH	+?	COU	690	32	Code 13; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6397Z		FRZBX	0233	29	M	WH	+?	COU	690	33	Code 12; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6396Z		FRZBX	0233	27	F	WH	+?	COU	690	34	Code 1087; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6393Z		FRZBX	0233	25	M	WH	+?		COU	690	35	Code 1088; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6386Z		FRZBX	0233	17	M	WH	+?		COU	690	36	Code 1091; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6392Z		FRZBX	0233	24	F	WH	+?		COU	690	37	Code 1089; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6394Z		FRZBX	0233	20	F	WH	+?		COU	690	38	Code 1090; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6403Z		FRZBX	0233	16	M	WH	+?		COU	690	39	Code 1092; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6470Z		FRZBX	0233	14	M	WH	+?		COU	690	40	Code 1093; unaffected; last examined on 3/11/83; parents are GM4707A and GM4711
GM6391Z		FRZBX	0233	12	F	WH	+?		COU	690	41	Code 1094; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM6395Z		FRZBX	0233	10	M	WH	+?		COU	690	42	Code 1095; unaffected; last examined on 3/8/83; parents are GM4707A and GM4711
GM5691Z		FRZBX	0233	60	F	WH	+?		MAT COU	690	43	Code 1083; unaffected; last examined on 4/4/82; sister of GM6680Z; mother of GM4707A; see GM5690A Lymphoid
GM6628Z		FRZBX	0233		F	WH	+?		MAT COU	690	44	Code 1082; unaffected; last examined on 3/26/83; sister of GM6680Z
GM6498Z		FRZBX	0233	35	M	WH	+?		MAT COU	690	45	Code 188; unaffected; last examined on 3/14/83; affected sibs are GM4877, GM4478, and GM4887; see GM6499 Lymphoid
GM6760Z		FRZBX	0233	9	F	WH	+?		MAT COU	690	46	Code 2275; unaffected; last examined on 4/2/83; father is GM6498Z
GM6749Z		FRZBX	0233	20	F	WH	+?		MAT COU	690	47	Code 2269; unaffected; last examined on 4/2/83; father is GM6498Z
GM6757Z		FRZBX	0233	14	M	WH	+?		MAT COU	690	48	Code 2272; unaffected; last examined on 4/2/83; father is GM6498Z
GM6753Z		FRZBX	0233	11	M	WH	+?		MAT COU	690	49	Code 2274; unaffected; last examined on 4/2/83; father is GM6498Z
GM6737Z		FRZBX	0233	18	M	WH	+?		MAT COU	690	50	Code 2271; unaffected; last examined on 4/2/83; father is GM6498Z

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			DISORDERS OF THE NERVOUS SYSTEM									
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM4877	3	EEMEM 20U	0233	34	F	WH	+-		AFF MAT COU	690	51	Code 189; clinically affected; onset at age 29 yrs; affected sibs are GM4887 & GM4478; see GM4876A Lymphoid
GM4865	4	EEMEM 20U	0233	43	M	WH	+?		MAT COU	690	52	Code 190; unaffected; last examined on 4/4/81; spouse of GM4877; mother is GM5559; see GM4864A Lymphoid
GM6503Z		FRZBX	0233	23	F	WH	+?		MAT COU	690	53	Code 1080; unaffected; last examined on 3/14/83; affected mother is GM4877
GM6502Z		FRZBX	0233	22	M	WH	+?		MAT COU	690	54	Code 1081; unaffected; last examined on 3/14/83; affected mother is GM4877
GM6523Z		FRZBX	0233	11	F	WH	+?		MAT COU	690	55	Code 1541; unaffected; last examined on 3/14/83; affected mother is GM4877
GM6504Z		FRZBX	0233	8	M	WH	+?		MAT COU	690	56	Code 1542; unaffected; last examined on 3/14/83; affected mother is GM4877
GM4717	2	EEMEM 20U	0233	53	F	WH	+?		MAT COU	690	57	Code 119; unaffected; last examined on 3/23/81; affected sibs are GM4877, GM4887, & GM4478; see GM4718A Lymphoid
GM6411Z		FRZBX	0233	55	M	WH	++		COU	690	58	Code 120; unaffected spouse of GM4717
GM6695Z		FRZBX	0233	18	M	WH	+?		MAT COU	690	59	Code 121; unaffected; last examined on 3/29/83; parents are GM4717 and GM6411Z
GM6406Z		FRZBX	0233	17	M	WH	+?		MAT COU	690	60	Code 2116; unaffected; last examined on 3/8/83; parents are GM4717 & GM6411Z
GM6402Z		FRZBX	0233	16	F	WH	+?		MAT COU	690	61	Code 2117; unaffected; last examined on 3/8/83; parents are GM4717 & GM6411Z
GM6414Z		FRZBX	0233	13	F	WH	+?		MAT COU	690	62	Code 2120; unaffected; last examined on 3/8/83; parents are GM4717 & GM6411Z
GM6401Z		FRZBX	0233	22	F	WH	+?		MAT COU	690	63	Code 122; unaffected; last examined on 3/8/83; parents are GM4717 & GM6411Z
*GM4887	2	EEMEM 20U	0233	48	F	WH	+-		AFF MAT COU	690	64	Code 184; clinically affected; affected sibs are GM4877 & GM4478; see GM4886A Lymphoid
GM6521Z		FRZBX	0233	26	F	WH	+?		MAT COU	690	66	Code 1549; unaffectud; last examined on 3/14/83; affected mother is GM4887

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM6507Z		FRZBX	0233	25	F	WH	+?	MAT COU	690	67	Code 1554; unaffected; last examined on 3/14/83; affected mother is GM4887
GM6500Z		FRZBX	0233	24	M	WH	+?	MAT COU	690	68	Code 1560; unaffected; last examined on 3/14/83; affected mother is GM4887
GM5535Z		FRZBX	0233	22	M	WH	+?	MAT COU	690	69	Code 1561; unaffected; last examined on 3/19/82; affected mother is GM4887; see GM5534A Lymphoid
GM4861	2	EEMEM 20U	0233	19	M	WH	+?	MAT COU	690	71	Code 187; unaffected; last examined on 4/2/81; affected mother is GM4887; see GM4860A Lymphoid
GM5527Z		FRZBX	0233	16	M	WH	+?	MAT COU	690	72	Code 1564; unaffected; last examined on 3/19/82; affected mother is GM4887; see GM5526A Lymphoid
GM6522Z		FRZBX	0233	16	F	WH	+?	MAT COU	690	73	Code 1565; unaffected; last examined on 3/14/83; affected mother is GM4887
GM4478	4	EEMEM 20U	0233	52	F	WH	+-	AFF MAT COU	690	77	Code 9; clinically affected; affected sibs are GM4887 & GM4877; 2 affected children and 4 with probable HD; see GM4479 Lymphoid
GM4476	2	EEMEM 20U	0233	58	M	WH	+-	AFF MAT COU	690	78	Code 8; clinically affected; onset at age 41 yrs; affected spouse is GM4478; 2 affected children and 4 with probable HD; neuro exam 3/82 shows difficulty in ambulation with frequent falls; see GM4477 Lymphoid
GM4845	3	EEMEM 20U	0233	37	F	WH	+?	MAT COU	690	79	Code 134; clinically unaffected; aff. parents are GM4478 & GM4476; 2 aff. sibs and 4 with probable HD; unaff. spouse is GM6552Z; neuro exam 3/82 shows brisk jaw jerk, nystagmus, & rigidity; see GM4844A Lymphoid
GM6552Z		FRZBX	0233	43	M	WH	++	IN-LAW	690	80	Code 135; clinically unaffected; unaffected spouse is GM4845
GM6505Z		FRZBX	0233	20	M	WH	+?	MAT COU	690	81	Code 136; unaffected; last examined on 3/14/83; unaffected mother is GM4845
GM6510Z		FRZBX	0233	17	F	WH	+?	MAT COU	690	82	Code 142; unaffected; last examined on 3/14/83; unaffected mother is GM4845

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6550Z		FRZBX	0233	15	M	WH	+?		MAT COU	690	83	Code 143; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM6549Z		FRZBX	0233	23	M	WH	+?		MAT COU	690	84	Code 144; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM6548Z		FRZBX	0233	12	F	WH	+?		MAT COU	690	85	Code 145; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM6542Z		FRZBX	0233	9	F	WH	+?		MAT COU	690	86	Code 147; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM6551Z		FRZBX	0233	8	M	WH	+?		MAT COU	690	87	Code 148; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM6543Z		FRZBX	0233	22	F	WH	+?		MAT COU	690	88	Code 137; unaffected; last examined on 3/21/83; unaffected mother is GM4845
GM4847	3	EEMEM 20U	0233	31	M	WH	+?		MAT COU	690	89	Code 149; unaffected; last examined on 4/2/81; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM4946A Lymphoid
GM6553Z		FRZBX	0233	27	F	WH	++		IN-LAW	690	90	Code 150; unaffected spouse of GM4847
GM6547Z		FRZBX	0233	11	F	WH	+?		MAT COU	690	91	Code 151; unaffected; last examined on 3/21/83; parents are GM4847 and GM6553Z
GM6545Z		FRZBX	0233	10	M	WH	+?		MAT COU	690	92	Code 152; unaffected; last examined on 3/21/83; parents are GM4847 and GM6553Z
GM6546Z		FRZBX	0233	9	F	WH	+?		MAT COU	690	93	Code 153; unaffected; last examined on 3/21/83; parents are GM4847 and GM6553Z
GM4689	2	EEMEM 20U	0233	30	F	WH	+-?		MAT COU	690	94	Code 154; probable HD; last examined on 3/23/81; 2 affected sibs and 3 with probable HD; affected parents are GM4478 and GM4476; see GM4690A Lymphoid
GM6467Z		FRZBX	0233	38	M	WH	++		IN-LAW	690	95	Code 155; unaffected spouse of GM4689
GM6620Z		FRZBX	0233	20	M	WH	+?		MAT COU	690	96	Code 156; unaffected; last examined on 3/24/83; parents are GM4689 & GM6467Z
GM6461Z		FRZBX	0233	19	F	WH	+?		MAT COU	690	97	Code 157; unaffected; last examined on 3/11/83; parents are GM4689 & GM6467Z
GM6468Z		FRZBX	0233	17	F	WH	+?		MAT COU	690	98	Code 158; unaffected; last examined on 3/11/83; parents are GM4689 & GM6467Z

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6404Z		FRZBX	0233	16	F	WH	+?		MAT COU	690	99	Code 159; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM6410Z		FRZBX	0233		F	WH	+?		MAT COU	691	1	Code 160; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM6472Z		FRZBX	0233	12	F	WH	+?		MAT COU	691	2	Code 161; unaffected; last examined on 3/11/83; parents are GM4689 & GM6467Z
GM6407Z		FRZBX	0233		M	WH	+?		MAT COU	691	3	Code 162; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM6405Z		FRZBX	0233		F	WH	+?		MAT COU	691	4	Code 163; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM6399Z		FRZBX	0233		F	WH	+?		MAT COU	691	5	Code 164; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM6409Z		FRZBX	0233		M	WH	+?		MAT COU	691	6	Code 165; unaffected; last examined on 3/8/83; parents are GM4689 & GM6467Z
GM4849	2	EEMEM 20U	0233	28	F	WH	+-		AFF MAT COU	691	7	Code 166; clinically affected; onset at age 24 yrs; affected parents are GM4478 & GM4476; 2 affected sibs and 4 with probable HD; neuro exam 3/82 shows rigidity with reinforcement; see GM5542B Lymphoid
GM6728Z		FRZBX	0233	28	M	WH	++		IN-LAW	691	8	Code 167; unaffected spouse of GM4849
GM4482	4	EEMEM 20U	0233	26	F	WH	+?		MAT COU	691	9	Code 123; unaffected; last examined on 11/22/80; 2 affected sibs and 4 with probable HD; affected parents are GM4478 & GM4476; see GM4289 Lymphoid
GM4857	2	EEMEM 20U	0233	24	F	WH	+-		AFF MAT COU	691	10	Code 170; clinically affected; onset at age 24 yrs; 1 affected sib and 4 with probable HD; affected parents are GM4478 & GM4476; neuro exam 3/82 shows bradykinesia; see GM4856A Lymphoid
GM6539Z		FRZBX	0233		M	WH	++		IN-LAW	691	11	Code 171; unaffected spouse of GM4857; see GM6538 Lymphoid
GM6531Z		FRZBX	0233	13	F	WH	+?		MAT COU	691	12	Code 1068; unaffected; last examined on 3/21/83; affected mother is GM4857; see GM6530 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM									
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6529Z		FRZBX	0233	11	M	WH	+?		MAT COU	691	13	Code 1069; unaffected; last examined on 3/21/83; affected mother is GM4857; see GM6528 Lymphoid
GM6533Z		FRZBX	0233	8	F	WH	+?		MAT COU	691	14	Code 1070; unaffected; last examined on 3/21/83; affected mother is GM4857; see GM6532 Lymphoid
GM4831	3	EEMEM 20U	0233	23	M	WH	+-?		MAT COU	691	16	Code 172; probable HD; affected parents are GM4478 & GM4476; 2 affected sibs & 3 with probable HD; neuro exam 3/82 showed a history of symptoms but no neurological signs; see GM4830A Lymphoid
GM4837	3	EEMEM 20U	0233	23	M	WH	+?		MAT COU	691	17	Code 173; unaffected; last examined on 2/3/82; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM5376 Lymphoid
GM4829	3	EEMEM 20U	0233	21	F	WH	+-?		MAT COU	691	18	Code 174; probable HD; affected parents are GM4478 & GM4476; 2 affected sibs & 3 with probable HD; neuro exam 3/82 shows mild left sided hyperreflexia & decreased eyelash burying on right; see GM4828A Lymphoid
GM6512Z		FRZBX	0233		M	WH	++		COU	691	19	Code 175; unaffected spouse of GM4829
GM4859	3	EEMEM 20U	0233	20	M	WH	+?		MAT COU	691	20	Code 178; unaffected; last examined on 4/2/81; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM4858A Lymphoid
GM6541Z		FRZBX	0233	21	F	WH	+?		MAT COU	691	21	Code 179; unaffected; last examined on 3/21/83; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM5536B Lymphoid
GM5551Z		FRZBX	0233	18	F	WH	+?		MAT COU	691	22	Code 180; unaffected; last examined on 3/19/82; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM4290 Lymphoid
GM4853	2	EEMEM 20U	0233	15	M	WH	+?		MAT COU	691	23	Code 182; unaffected; last examined on 4/2/81; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM4852A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4855	2	EEMEM 20U	0233	12	M	WH	+?	MAT COU	691	24	Code 183; unaffected; last examined on 4/2/81; affected parents are GM4478 and GM4476; 2 affected sibs and 4 with probable HD; see GM4854A Lymphoid
GM6630Z		FRZBX	0233	86	M	WH	+?	MAT GR UNC	691	25	Code 1040; unaffected; last examined on 3/26/83; probable escapee; mother had HD; see GM6629 Lymphoid
GM5699Z		FRZBX	0233	61	F	WH	+?	PAT COU	691	26	Code 328; unaffected; last examined on 4/6/82; at risk father died at age 40; see GM5700A Lymphoid
GM6422Z		FRZBX	0233	56	M	WH	+?	PAT COU	691	27	Code 1483; unaffected; last examined on 3/11/83; at risk father died at age 40; see GM6421 Lymphoid
GM6735Z		FRZBX	0233		M	WH	+?	MAT COU	691	28	Code 1493; unaffected; last examined on 4/2/83; at risk father died at age 40
GM5649Z		FRZBX	0233	105	F	WH	+?	MAT COU	691	29	Code 329; unaffected; last examined on 3/26/82; probable escapee; 9 affected sibs
GM5679Z		FRZBX	0233	58	M	WH	+-	AFF MAT COU	691	30	Code 356; clinically affected; affected sister is GM5609Z; see GM5678A Lymphoid
GM5609Z		FRZBX	0233	52	F	WH	+-	AFF MAT COU	691	31	Code 344; clinically affected; affected brother is GM5679Z; see GM5610A Lymphoid
*GM4801	3	EEMEM 20U	0233	45	F	WH	+?	MAT COU	691	32	Code 355; unaffected; last examined on 3/27/81; affected sibs are GM5679Z & GM5609Z; see GM4800A Lymphoid
GM4809	2	EEMEM 20U	0233	42	M	WH	+?	MAT COU	691	33	Code 354; unaffected; last examined on 3/27/81; affected sibs are GM5679Z & GM5609Z; see GM4808A Lymphoid
GM6456Z		FRZBX	0233	79	F	WH	+?	MAT COU	691	34	Code 360; unaffected; last examined on 3/11/83; probable escapee; 9 affected sibs; sister is GM5649Z
GM5685Z		FRZBX	0233	72	F	WH	++	IN-LAW	691	35	Code 373; unaffected spouse of HD patient; affected children are GM5683Z and GM5669Z; see GM5684B Lymphoid
GM5683Z		FRZBX	0233	49	F	WH	+-	AFF MAT COU	691	36	Code 374; clinically affected; father had HD; mother is GM5685Z; affected brother is GM5669Z; see GM5682A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM5661Z		FRZBX	0233		F	WH	+?		MAT COU	691	37	Code 378; unaffected; last examined on 4/4/82; affected sibs are GM5683Z and GM5669Z; see GM5660A Lymphoid
GM5669Z		FRZBX	0233	46	M	WH	+-		AFF MAT COU	691	38	Code 379; clinically affected; affected sister is GM5683Z; see GM5668A Lymphoid
GM6375Z		FRZBX	0233	57	F	WH	+?		MAT COU	691	39	Code 385; unaffected; last examined on 3/8/83; spouse of HD patient; mother of GM6369Z; see GM6376 Lymphoid
GM4819	2	EEMEM 20U	0233	48	M	WH	+-		AFF MAT COU	691	40	Code 387; clinically affected; affected son is GM5625Z; affected sibs are GM5555Z and GM4727; neuro exam 3/82 shows moderate chorea with parakinesias; see GM6348 Lymphoid
GM5641Z		FRZBX	0233	41	F	WH	++		COU	691	41	Code 388; unaffected spouse of GM4819; affected son is GM5625Z; see GM5642A Lymphoid
GM5625Z		FRZBX	0233	25	M	WH	+-		AFF MAT COU	691	42	Code 389; clinically affected; affected father is GM4819; see GM5626A Lymphoid
GM6444Z		FRZBX	0233	44	F	WH	+-		AFF MAT COU	691	43	Code 411; clinically affected; last examined 3/83; affected sibs are GM4819, GM5555Z, & GM4727; see GM6443 Lymphoid
GM5555Z		FRZBX	0233		F	WH	+-		AFF MAT COU	691	44	Code 396; clinically affected; affected sibs are GM4819, GM6444Z, and GM4727; see GM5554B Lymphoid
GM4727	3	EEMEM 20U	0233	40	M	WH	+-?		MAT COU	691	45	Code 393; probable HD; affected sibs are GM4819, GM5555Z, and GM6444Z
GM5605Z		FRZBX	0233	37	F	WH	+?		MAT COU	691	46	Code 404; unaffected; last examined on 3/26/82; affected sibs are GM4819, GM6444Z, GM5555Z, & GM4727; see GM5606A Lymphoid
GM4693	5	EEMEM 20U	0233	33	M	WH	+?		MAT COU	691	47	Code 414; unaffected; last examined on 3/23/81; affected sibs are GM6444Z, GM5555Z, GM4819, & GM4727; see GM4694 Lymphoid
GM5599Z		FRZBX	0233	76	M	WH	++		IN-LAW	691	48	Code 419; unaffected spouse of HD patient; affected children are GM4799, GM5621Z, & GM5613Z; see GM5600A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
			Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)								
GM4797	2	EEMEM 20U	0233	50	F	WH	+?	MAT COU	691	49	Code 425; unaffected; last examined on 3/27/81; affected sibs are GM4799, GM5621Z, & GM5613Z; see GM4796 Lymphoid
*GM4799	2	EEMEM 20U	0233	48	M	WH	+-	AFF MAT COU	691	50	Code 420; clinically affected; affected sibs are GM5621Z & GM5613Z; neuro exam 3/82 shows flexion of distal interphalangeal joint of thumb & no vertical optokinetic response; see GM4798A Lymphoid
GM5723Z		FRZBX	0233	46	M	WH	+?	MAT COU	691	51	Code 423; unaffected; last examined on 4/6/82; affected sibs are GM5621Z, GM4799, & GM5613Z; see GM5724A Lymphoid
GM4795	2	EEMEM 20U	0233	42	F	WH	+?	MAT COU	691	52	Code 426; unaffected; last examined on 3/27/81; affected sibs are GM5621Z, GM4799, & GM5613Z; see GM4794A Lymphoid
GM5621Z		FRZBX	0233	41	M	WH	+-	AFF MAT COU	691	53	Code 424; clinically affected; affected sibs are GM4799 & GM5613Z; see GM5622A Lymphoid
GM5613Z		FRZBX	0233	39	F	WH	+-	AFF MAT COU	691	54	Code 534; clinically affected; affected sibs are GM5621Z & GM4799; see GM5614A Lymphoid
GM5739Z		FRZBX	0233	36	M	WH	+?	MAT COU	691	55	Code 1248; unaffected; last examined on 4/6/82; affected sibs are GM5621Z, GM4799, & GM5613Z; see GM5740A Lymphoid
GM5639Z		FRZBX	0233	34	M	WH	+?	MAT COU	691	56	Code 1249; unaffected; last examined on 3/26/82; affected sibs are GM4799, GM5621Z, & GM5613Z; see GM5640A Lymphoid
GM5744Z		FRZBX	0233	72	M	WH	+?	MAT COU	691	57	Code 429; unaffected; last examined on 4/82; 9 affected sibs; sister is GM5649Z
GM4787	4	EEMEM 20U	0233	54	F	WH	++	COU	691	58	Code 460; clinically unaffected spouse of HD patient; affected children are GM4715, GM4709, & GM4687; see GM4786A Lymphoid
GM4715	3	EEMEM 20U	0233	40	M	WH	+-	AFF MAT COU	691	59	Code 474; clinically affected; age of onset 19 yrs; affected sibs are GM4709 & GM4687; neuro exam 3/82 shows slight chorea in toes, fingers, & shoulders; seems more demented than motor impaired; see GM4716A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4709	2	EEMEM 20U	0233	38	F	WH	+-	AFF MAT COU	691	60	Code 463; clinically affected; onset at age 34 yrs; affected sibs are GM4715 & GM4687; 1st cousin once removed to proband; neuro exam 3/82 shows dyskinesia & impaired gaze; see GM4710A Lymphoid
GM4687	3	EEMEM 20U	0233	38	F	WH	+-	AFF MAT COU	691	61	Code 469; clinically affected; onset at age 34 yrs; affected sibs are GM4715 & GM4709; neuro exam 3/82 shows mild chorea, marginal dystonia, hypomimia, and bradykinesia; see GM4688A Lymphoid
GM6682Z		FRZBX	0233		M	WH	++	COU	691	62	Code 480; unaffected spouse of HD patient
GM5579Z		FRZBX	0233	45	M	WH	+?	MAT COU	691	63	Code 482; unaffected; last examined on 3/26/82; mother had HD; 1st cousin once removed to proband; see GM5580A Lymphoid
GM5607Z		FRZBX	0233	54	F	WH	+-	AFF COU	691	64	Code 526; affected; onset at age 47 yrs; both parents were affected; affected son is GM5627Z; neuro exam 3/82 shows prominent chorea, moderate dystonia, & some dementia; see GM5608A Lymphoid
GM5593Z		FRZBX	0233	72	M	WH	++	IN-LAW	691	65	Code 527; unaffected spouse of GM5607Z; affected son is GM5627Z; see GM5594A Lymphoid
GM5627Z		FRZBX	0233	36	M	WH	+-	AFF COU	691	66	Code 859; clinically affected; affected mother is GM5607Z; see GM5628A Lymphoid
GM5557Z		FRZBX	0233	72	F	WH	+?	MAT COU	691	67	Code 15; unaffected; last examined on 3/21/82; 2 sibs had HD; paternal aunt of GM5607Z
GM5547Z		FRZBX	0233	62	M	WH	+?	MAT COU	691	68	Code 7; unaffected; last examined on 3/19/82; 2 sibs had HD; paternal uncle of GM5607Z; see GM5546B Lymphoid
GM5689Z		FRZBX	0233	42	M	WH	+-	AFF PAT COU	691	69	Code 1100; clinically affected; both parents were affected; affected sib is GM4869; 1st cousin to proband; see GM5688A Lymphoid
GM4869	2	EEMEM 20U	0233	30	F	WH	+-	AFF PAT COU	691	70	Code 558; clinically affected; onset at age 29 yrs; both parents affected; affected sib is GM5689Z; 1st cousin to proband; see GM4868A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM6436Z		FRZBX	0233	71	F	WH	+?	MAT COU	691	71	Code 67; unaffected; last examined on 3/11/83; one affected sib; spouse of HD patient; affected daughter is GM4767; see GM6437 Lymphoid
GM4767	2	EEMEM 20U	0233	42	F	WH	+-	AFF MAT COU	691	72	Code 69; clinically affected; spouse is GM6684Z; affected daughter is GM6596Z; neuro exam 3/82 shows prominent chorea of all extremities & reduced saccades - vertical > horizontal; see GM4766A Lymphoid
GM5603Z		FRZBX	0233	37	M	WH	+?	MAT COU	691	73	Code 72; unaffected; last examined on 3/26/82; affected sib is GM4767; see GM5604A Lymphoid
GM5571Z		FRZBX	0233	34	F	WH	+?	MAT COU	691	74	Code 73; unaffected; last examined on 3/26/82; affected sib is GM4767; see GM5572A Lymphoid
GM6780Z		FRZBX	0233	34	M	WH	+?	MAT COU	691	75	Code 76; unaffected; last examined on 4/4/83; affected sib is GM4767
GM6596Z		FRZBX	0233	40	F	WH	+-?	1ST COU	691	76	Code 1158; probable HD; affected mother is GM4767; father is GM6684Z; see GM6595 Lymphoid
GM5559	4	EEMEM 20U	0233	68	F	WH	+?	MAT COU	691	77	Code 555; unaffected; last examined on 3/82; 1 affected sib; sister is GM6438Z; son is GM4865; see GM5558A Lymphoid
GM4815	2	EEMEM 20U	0233	74	M	WH	++	IN-LAW	691	78	Code 634; unaffected spouse of HD patient; affected children are GM4287 and GM4285; 4 other affected children are deceased; see GM4814A Lymphoid
*GM4793	3	EEMEM 20U	0233	52	M	WH	+?	MAT COU	691	79	Code 640; unaffected; last examined on 3/27/81; affected sibs are GM4287 & GM4285; 4 others are deceased; father is GM4815; see GM4792A Lymphoid
GM4287	2	EEMEM 20U	0233	44	M	WH	+-	AFF MAT COU	691	81	Code 647; clinically affected; onset at age 30 yrs; affected sib is GM4285; affected dau. is GM4723; father is GM4815; neuro exam 3/82 shows bradykinesis, Parkinsonian features, & mild chorea; PD gait & dystonia; see GM4288 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4723	2	EEMEM 20U	0233	19	F	WH	+-	AFF MAT COU	691	82	Code 653; clinically affected; affected father is GM4287; neuro exam 3/82 shows bradykinesis, juvenile features of HD with markedly reduced saccadic velocity, Parkinsonian features, & marginal dystonia; see GM4724 Lymphoid
GM4285	2	EEMEM 20U	0233	41	M	WH	+-	AFF MAT COU	691	83	Code 659; clinically affected; onset at age 30 yrs; affected sib is GM4287; 4 others are deceased; father is GM4815; neuro exam 3/82 shows bradykinesis, dystonia, & chorea; see GM4286 Lymphoid
*GM4729	2	EEMEM 20U	0233	39	F	WH	+?	MAT COU	691	84	Code 665; unaffected; last examined on 3/27/81; affected sibs are GM4287 & GM4285; 4 others are deceased; father is GM4815; see GM4730A Lymphoid
GM4791	3	EEMEM 20U	0233	101	F	WH	+?	COU	691	85	Code 663; unaffected; last examined on 3/27/81; probable escapee; spouse of HD patient; see GM4790A Lymphoid
GM4735	3	EEMEM 20U	0233	62	M	WH	+?	MAT COU	691	86	Code 560; unaffected; last examined on 3/23/81; son of GM4791; affected spouse is GM5562Z; affected daughter is GM4721; see GM4736A Lymphoid
GM5562Z		FRZBX	0233	60	F	WH	+-	AFF PAT AUNT	691	87	Code 559; clinically affected; spouse is GM4735; affected daughter is GM4721; neuro exam 3/82 shows inability to talk, ability to walk 4-5 steps only with assistance; terminal stages of HD; see GM4818B Lymphoid
*GM4721	3	EEMEM 20U	0233	37	F	WH	+-	AFF PAT COU	691	88	Code 561; clinically affected; parents are GM4735 and GM5562Z; neuro exam 3/82 shows chorea & facial twitches; 1st cousin to proband; see GM4722A Lymphoid
GM6776Z		FRZBX	0233	76	M	WH	+?	MAT COU	691	89	Code 917; unaffected; last examined on 4/4/83; affected sib is GM6766Z; father had HD; see GM6777 Lymphoid
GM6634Z		FRZBX	0233	51	M	WH	+?	MAT COU	691	90	Code 920; unaffected; last examined on 3/26/83; affected sib is GM6766Z; see GM6633 Lymphoid
GM6656Z		FRZBX	0233	63	F	WH	+?	MAT COU	691	91	Code 923; unaffected; last examined on 3/26/83; affected sib is GM6766Z; see GM6655A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM6766Z		FRZBX	0233	61	M	WH	+-	AFF MAT COU	691	92	Code 929; clinically affected; affected children are GM6652Z & GM6632Z; unaffected spouse is GM6615Z; see GM6767 Lymphoid
GM6615Z		FRZBX	0233	51	F	WH	++	IN-LAW	691	93	Code 930; unaffected spouse of GM6766Z; affected children are GM6652Z & GM6632Z
GM6652Z		FRZBX	0233	45	M	WH	+-?	MAT COU	691	94	Code 931; probable HD; affected sib is GM6632Z; parents are GM6766Z & GM6615Z; see GM6651A Lymphoid
GM6632Z		FRZBX	0233	41	M	WH	+-	AFF MAT COU	691	95	Code 934; clinically affected; affected sib is GM6652Z; parents are GM6766Z & GM6615Z; see GM6631 Lymphoid
GM6594Z		FRZBX	0233	55	M	WH	+?	MAT COU	691	96	Code 940; unaffected; last examined on 3/24/83; affected sibs are GM5427Z, GM6567Z, and GM6448Z; see GM6593A Lymphoid
GM5427Z		FRZBX	0233	44	F	WH	+-	AFF MAT COU	691	98	Code 963; clinically affected; affected sibs are GM6567Z and GM6448Z; see GM5428 Lymphoid
GM6586Z		FRZBX	0233	43	M	WH	+?	MAT COU	691	99	Code 994; unaffected; last examined on 3/24/83; affected sibs are GM5427Z, GM6567Z, & GM6448Z; see GM6585A Lymphoid
GM6567Z		FRZBX	0233	37	F	WH	+-	AFF MAT COU	692	1	Code 1002; clinically affected; affected sibs are GM5427Z and GM6448Z; see GM6566 Lymphoid
GM6448Z		FRZBX	0233	37	F	WH	+-?	MAT COU	692	2	Code 932; probable HD; affected sibs are GM5427Z and GM6567Z; affected spouse is GM6652Z; see GM6447 Lymphoid
GM6424Z		FRZBX	0233	19	F	WH	+?	MAT COU	692	3	Code 2992; unaffected; last examined on 3/11/83; affected parents are GM6448Z & GM6652Z; see GM6423 Lymphoid
GM6450Z		FRZBX	0233	16	F	WH	+?	MAT COU	692	4	Code 2996; unaffected; last examined on 3/11/83; affected parents are GM6448Z & GM6652Z; see GM6449 Lymphoid
GM6446Z		FRZBX	0233	14	F	WH	+?	MAT COU	692	5	Code 2999; unaffected; last examined on 3/11/83; affected parents are GM6448Z & GM6652Z; see GM6445 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMD	FAMM	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM6602Z		FRZBX	0233	18	F	WH	+?	MAT COU	692	6	Code 2993; unaffected; last examined on 3/24/83; affected parents are GM6448Z & GM6652Z; see GM6601 Lymphoid
GM6644Z		FRZBX	0233	23	M	WH	+?	MAT COU	692	7	Code 933; unaffected; last examined on 3/26/83; affected parents are GM6448Z & GM6652Z; see GM6643 Lymphoid
GM5485Z		FRZBX	0233	33	M	WH	+?	MAT COU	692	8	Code 1010; unaffected; last examined on 3/15/82; affected sibs are GM5427Z, GM6567Z, & GM6448Z; see GM5486A Lymphoid
GM6582Z		FRZBX	0233	29	F	WH	+-	AFF COU	692	9	Code 950; clinically affected; niece of GM5427Z; see GM6581 Lymphoid
GM5447Z		FRZBX	0233	46	M	WH	+?	PAT 1ST COU	692	10	Code 533; unaffected; last examined on 3/15/82; affected spouse is GM5613Z; 1st cousin once removed to proband; see GM5448 Lymphoid
GM5451Z		FRZBX	0233	44	F	WH	+?	PAT 1ST COU	692	11	Code 537; unaffected; last examined on 3/15/82; 1st cousin once removed to proband; see GM5452A Lymphoid
GM5453Z		FRZBX	0233	38	F	WH	+?	PAT 1ST COU	692	12	Code 543; unaffected; last examined on 3/15/82; 1st cousin once removed to proband; see GM5454A Lymphoid
GM5475Z		FRZBX	0233	35	M	WH	+?	PAT 1ST COU	692	13	Code 547; unaffected; last examined on 3/11/83; 1st cousin once removed to proband; see GM5476A Lymphoid
GM6480Z		FRZBX	0233	72	M	WH	+?	PAT UNC	692	14	Code 552; unaffected; last examined on 3/14/83; 5 affected sibs; brother of GM4867; see GM6481 Lymphoid
GM6363Z		FRZBX	0233	59	F	WH	+?	PAT AUNT	692	15	Code 585; unaffected; last examined on 3/8/83; 5 affected sibs; brother is GM4867; see GM6364 Lymphoid
GM4745	2	EEMEM 20U	0233	57	F	WH	+?	PAT AUNT	692	16	Code 586; unaffected; last examined on 3/23/81; 5 affected sibs; brother is GM4867; see GM4734A Lymphoid
*GM4779	2	EEMEM 20U	0233	52	M	WH	+-	AFF PAT UNC	692	17	Code 587; clinically affected; four affected sibs; brother of GM4867; affected children are GM4691 & GM6440Z; unaffected spouse is GM4775; neuro exam 3/82 shows chorea, poor speech, & impaired swallowing

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
*GM4775	2	EEMEM 20U	0233	46	F	WH	++		COU	692	I8	Code 588; unaffected spouse of GM4779; affected children are GM4691 and GM6440Z
GM4691	4	EEMEM 20U	0233	31	M	WH	+-		AFF 1ST COU	692	19	Code 589; clinically affected; onset at age 25 yrs; affected sib is GM6440Z; parents are GM4779 & GM4775; neuro exam 3/82 shows prominent laughing (? pseudobulba) & involuntary vocalizations; dementia & dystonia; see GM4692A Lymphoid
GM6440Z		FRZBX	0233	29	M	WH	+-		AFF 1ST COU	692	20	Code 591; clinically affected; affected sib is GM4691; parents are GM4779 and GM4775; neuro exam 3/82 shows reduced vertical > horizontal saccades, mild dystonia, & prominent chorea; see GM6439 Lymphoid
GM6772Z		FRZBX	0233	37	M	WH	+-		AFF PAT UNC	692	21	Code 598; clinically affected; last examined on 4/4/83; four affected sibs; brother of GM4867; see GM6773 Lymphoid
GM6676Z		FRZBX	0233	47	M	WH	+?		PAT UNC	692	22	Code 603; unaffected; last examined on 3/29/83; 5 affected sibs; brother is GM4867; see GM6677 Lymphoid
GM4781	2	EEMEM 20U	0233	71	M	WH	+?		PAT GR UNC	692	23	Code 624; unaffected; last examined on 3/27/81; 4 affected sibs; see GM4780A Lymphoid
Lissencephaly Syndrome - 24720												
GM6047	2	EEMEM 20U	0417	4MO	M	WH	--	A	PROBAND			Typical facies (bitemporal hollowing, thin downturned upper lip, micrognathia), umbilical hernia, sacral sinus tract, and cryptorchidism; 46,XY,r(17)(p13.3q25.3); unbalanced
GM6097A	3	EEMEM 20U	0417	15MO	F	WH	--	A	PROBAND			Typical facies (bitemporal hollowing, thin down turned upper lip, micrognathia, microcephaly, high arched palate); ventricular septal defect; 46,XX,-17,+der(17),t(7;17)(17qter>17p13.2::7p22.3>7pter)pat; unbalanced
Neuropathy, Congenital Sensory, with Anhidrosis - 25680												
GM2958	2	EEMEM 20U	0105	13	M	WH	--		PROBAND			Insensitivity to pain
Neuroaxonal Dystrophy, Infantile (Seitelberger) - 25660												
GM6324	5	EEMEM 20U	0448	42MO	M	WH	--		PROBAND	950	1	Portuguese-American; similarly affected brother; widespread neuronal loss and axonal degeneration with spheroids; cerebellar atrophy; progressive CNS degeneration involving primarily motor systems; normal lysosomal enzymes

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Neuroaxonal Dystrophy, Infantile (Seitelberger) - 25660											
GM6325	5	EEMEM 20U	0448	30	F	WH	+-(O)	MOTHER	950	2	Portuguese-American; clinically unaffected; two affected sons
Neuroblastoma - 25670											
*GM3320C	55	EEMEM 10I	0152	13MO	M	WH	--	PROBAND			IMR-32; ATCC CCL 127; fibroblast and neuroblast-like cell culture; G6PD type B; hyperdiploid; short arm of chromosome 1 shows an HSR
Olivopontocerebellar Atrophy - 16440, 50, 60, 70, and 25830											
GM3561	3	EEMEM 20U	0110	22	F	BL		PROBAND	240	1	Probably type III; see GM3562 Lymphoid; positive family history
*GM6927	2	EEMEM 20U	0473	29	M	WH	+-	PROBAND	972	1	Olivopontocerebellar atrophy type I; spinocerebellar ataxia; six affected generations; see GM6926 Lymphoid
Primary Affective Disorder - 12548 and 30920											
GM2790	5	EEMEM 20U	0137	64	M			PROBAND	276	1	Primary affective disease, depressed; stuporous for months; serious depression; family history of depression
GM5215	6	EEMEM 20U	0308	23	F	WH		PROBAND	802	1	Type I bipolar illness; cyclothymic personality; onset at age 18; alcoholic; drug abuse; eight episodes of major depression; six episodes of mania; on Lithium; skin biopsy (posterior iliac crest); affected mother is GM6139 Lymph.
GM5228	7	EEMEM 20U	0308	17	F	WH		AFF SIS	802	2	Probable anorexia nervosa; alcoholic; onset at age 14; skin biopsy (posterior iliac crest); affected mother is GM6139 Lymphoid
*GM5224	6	EEMEM 20U	0308	20	M	WH		PROBAND	811	1	Type I bipolar illness; onset at age 17; three manic episodes; drug abuse; on Lithium; positive family history; skin biopsy (posterior iliac crest); see GM4984 Lymphoid
*GM5225	6	EEMEM 20U	0308	28	M	WH		AFF BRO	811	2	Type I bipolar illness; onset at age 17; 5 major depressive episodes and 2 manic episodes; on Lithium; skin biopsy (posterior iliac crest); see GM4986 Lymphoid
*GM7199	2	EEMEM 20U	0308	16	M	WH		AFF MAT COU	811	7	Cyclothymic personality; minor depression; onset at age 14; see GM7200 Lymphoid; maternal first cousin of a bipolar male

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				DISORDERS OF THE NERVOUS SYSTEM							
Primary Affective Disorder - 12548 and 30920											
*GM7209	3	EEMEM 20U	0308	28	F	WH		AFF MAT COU	811	8	One episode of hypomania; onset at age 25; no hospitalization; no medication; maternal first cousin of a bipolar male; see GM7208 Lymphoid; sister of GM7199
*GM7211	1	EEMEM 20U	0308	26	F	WH		MAT COU	811	9	Clinically unaffected; see GM7210 Lymphoid; sister of GM7199; maternal first cousin of a bipolar male
*GM7269	1	EEMEM 20U	0308	30	F	WH		MAT COU	811	10	At risk; clinically unaffected; see GM7268 Lymphoid; paternal aunt & cousin have bipolar illness; sister of GM7199; maternal first cousin of a bipolar male
GM7393	3	EEMEM 20U	0308	48	F	WH		AFF MAT AUNT	811	11	Type II bipolar illness with major depression; onset at age 34; 2 episodes of major depression; 1 hypomanic episode; hospitalized; on Lithium and Elavil; see GM7392 Lymphoid
GM7413	5	EEMEM 20U	0308	59	F	WH		AUNT	811	12	Clinically unaffected; in-law of proband; mother of GM7199, GM7209, GM7211, and GM7269; wife of GM7424; see GM7411 Lymphoid
GM7424	2	EEMEM 20U	0308	63	M	WH		MAT UNCLE	811	13	Clinically unaffected; reduced at risk; husband of GM7413; father of GM7199, GM7209, GM7211, and GM7269; see GM7423 Lymphoid
GM5226	6	EEMEM 20U	0308	43	F	WH		PROBAND	812	1	Type I bipolar illness; positive family history; onset at age 18; 3 episodes of major depression, indeterminate number of manic episodes; on Lithium; skin biopsy (posterior iliac crest); see GM4985 Lymphoid
GM5218A	9	EEMEM 20U	0308	55	M	WH		AFF BRO	812	2	Schizo-affective manic and schizo-affective depressed; anti-social personality; onset at age 19; 4 episodes of major depression, one episode of mania, & 4 episodes of hypomania; on Lithium; skin bx. (post. iliac crest); see GM4946A Lymph.
GM7379	2	EEMEM 20U	0308	15	F	WH		DAU	812	6	At risk; mother has type I bipolar illness; see GM7378 Lymphoid
GM7240	2	EEMEM 20U	0308	21	F	WH		SIS	820	3	At risk; clinically unaffected; mother and brother have type I bipolar illness; see GM7239 Lymphoid; proband is GM5134 Lymphoid

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DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
GM7246	2	EEMEM 20U	0308	24	M	WH		AFF BRO	820	4	Alcoholism; minor depression; onset of alcoholism at age 15 and depression at age 23; mother and brother have type I bipolar illness; see GM7245 Lymphoid; proband is GM5134 Lymphoid
GM7398A	4	EEMEM 20U	0308	21	F	WH		DAU	823	3	At risk; clinically unaffected; mother, sister, & 2 brothers have type I bipolar illness; see GM6972 Lymphoid; proband is GM5156 Lymphoid
GM7321	5	EEMEM 20U	0308	50	M	WH		SPOUSE	823	4	Clinically unaffected; wife, daughter, & 2 sons have type I bipolar illness; proband is GM5156 Lymphoid; see GM6973 Lymphoid
GM7320	4	EEMEM 20U	0308	18	F	WH		DAU	823	5	At risk; clinically unaffected; mother, sister, & 2 brothers have type I bipolar illness; see GM6974 Lymphoid; proband is GM5156 Lymphoid
GM7219	4	EEMEM 20U	0308	28	F	WH		AFF DAU	823	6	Type I bipolar illness; hyperthymic personality; generalized anxiety; multiple episodes of depression; 3 episodes of mania; currently receiving Lithium, Haldol, Cogentin, & Thorazine; see GM7218 Lymphoid; proband is GM5156 Lymphoid
GM7317	3	EEMEM 20U	0308	53	F	WH		SIS	823	7	Clinically unaffected; at risk; sister has type I bipolar illness; see GM7316 Lymphoid; proband is GM5156 Lymphoid
GM7336	3	EEMEM 20U	0308	56	M	WH		BRO	823	9	Clinically unaffected; one episode of minor depression after receiving Butazolidine for arthritis; sister has bipolar manic-depressive illness; see GM7335A Lymphoid; proband is GM5156 Lymphoid
GM7395	4	EEMEM 20U	0308	26	M	WH		AFF SON	823	10	Type I bipolar illness; drug abuse; generalized anxiety disorder; anti-social behavior; onset at age 13; one manic episode lasting 6 weeks; hospitalized; on Haldol and Lithium; see GM7394 Lymphoid; proband is GM5156 Lymphoid
GM7551	5	EEMEM 20U	0308	40	F	WH		SIS	823	11	At risk; clinically unaffected sib of a type I bipolar female; see GM7550 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
Primary Affective Disorder - 12548 and 30920											
GM7781	2	EEMEM 20U	0308	62	M	WH		BRO	823	12	Clinically unaffected sib of a type I bipolar female; proband is GM5156 Lymphoid
GM5216	5	EEMEM 20U	0308	60	F	WH		PROBAND	830	1	Unipolar illness; positive family history; two episodes of dysphoria; poor appetite; loss of energy, loss of interest, & slowed down movements; onset at age 49; on Elavil; skin biopsy (post. iliac crest); see GM4972 Lymphoid
GM7153	2	EEMEM 20U	0308	27	M	WH		AFF NEPHEW	830	2	Probable unipolar illness; major depression; onset at age 17; required hospitalization & medication; son of GM7155; see GM7152 Lymphoid
GM7155	2	EEMEM 20U	0308	65	M	WH		AFF BRO	830	3	Type I bipolar illness; onset at age 18; indeterminate number of episodes of major depression; indeterminate number of manic episodes; no hospitalization; no medication; see GM7154 Lymphoid
GM7205	2	EEMEM 20U	0308	25	F	WH		AFF DAU	830	4	Type II bipolar illness; major depression; onset at age 15; 2 episodes of depression; 2 episodes of hypomania; treated with Elavil and Lithium; see GM7201 Lymphoid
GM7221	3	EEMEM 20U	0308	32	F	WH		DAU	830	5	At risk; see GM7220 Lymphoid; mother has unipolar illness, 1 sister has type II bipolar illness, & 1 sister has minor depression
GM7223B	7	EEMEM 20U	0308	30	F	WH		NIECE	830	6	At risk; father (GM7155) has type I bipolar illness; see GM7222 Lymphoid
GM7258	2	EEMEM 20U	0308	29	F	WH		AFF NIECE	830	7	Type I bipolar illness; onset at age 17; at least 10 episodes of major depression; 2 episodes of mania; hospitalizations; on Lithium, Mellaril, Haldol, Cogentin, Elavil, Thorazine, & Stelazine; see GM7257 Lymphoid
GM7325	3	EEMEM 20U	0308	60	F	WH		AFF SIS	830	8	Type I bipolar illness; mother of GM7258; see GM7324 Lymphoid
GM7329	6	EEMEM 20U	0308	72	M	WH		BRO	830	9	Clinically unaffected; see GM7328 Lymphoid; brother and sister have bipolar illness

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DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
GM7331	3	EEMEM 20U	0308	63	M	WH		BRO-IN-LAW	830	10	Alcoholism; husband of GM7325; father of GM7258; see GM7330 Lymphoid
GM7749	3	EEMEM 20U	0308	36	M	WH		SON	830	11	At risk; hyperthymic personality onset at age 13; mother has unipolar illness and a sister has type II bipolar illness; see GM7748 Lymphoid
GM7924	3	EEMEM 20U	0308	62	M	WH		SPOUSE	830	12	Clinically unaffected; wife has unipolar illness; one daughter has type II bipolar illness and another minor depression; see GM7800 Lymphoid
GM5219	6	EEMEM 20U	0308	30	F	WH		AFF SIS	832	1	Depressive personality; type II bipolar illness with major depression and generalized anxiety disorder; onset at age 11; 3 episodes of major depression & 1 episode of hypomania; pos. family history; skin biopsy (post. iliac crest)
GM5227	6	EEMEM 20U	0308	27	F	WH		PROBAND	832	2	Type I bipolar illness; positive family history; onset at age 22; 3 episodes of major depression; indeterminate number of episodes of mania; hyperthymic personality; on Lithium; skin biopsy (posterior iliac crest); see GM4996 Lymph.
GM5220	7	EEMEM 20U	0308	55	M	WH		PROBAND	833	1	Type I bipolar illness; positive family history; onset at age 38; three episodes of major depression; five episodes of mania; on Lithium; skin biopsy (posterior iliac crest); see GM4949B Lymphoid
GM5221	6	EEMEM 20U	0308	65	M	WH		PROBAND	834	1	Type II bipolar illness; positive family history; onset at age 43; 3 episodes of major depression; 3 episodes of hypomania; on Lithium; a daughter has acute intermittent porphyria; pt tested neg; see GM4952A Lymph.; skin bx (p. iliac crest)
GM5222	7	EEMEM 20U	0308	38	M	WH		PROBAND	835	1	Type I bipolar illness; positive family history; onset at age 32; hyperthymic personality; four episodes of mania; unknown number of minor depressive episodes; on Lithium; skin biopsy (posterior iliac crest); see GM4950A Lymphoid
GM5217	7	EEMEM 20U	0308	29	F	WH		AFF SIS	835	2	Type I bipolar illness; positive family history; onset at age 20; one manic episode and one major depressive episode; on Lithium and Dalmane; skin biopsy (posterior iliac crest)

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				DISORDERS OF THE NERVOUS SYSTEM							
Primary Affective Disorder - 12548 and 30920											
GM7242	2	EEMEM 20U	0308	18	M	WH		PROBAND	836	1	See GM7241 Lymphoid; at risk; mother has type II bipolar illness
GM7250	3	EEMEM 20U	0308	18	M	WH		TWIN	836	3	See GM7249 Lymphoid; at risk; dizygotic twin of GM7242; mother has type II bipolar illness
GM4931	6	EEMEM 20U	0308	43	M	WH		PROBAND	837	1	Bipolar illness; positive family history; onset at age 33; chronic major depression; on Protriptyline; skin biopsy (posterior iliac crest)
GM7244	2	EEMEM 20U	0308	24	F	WH		PROBAND	1010	1	At risk; clinically unaffected; see GM7243 Lymphoid; mother has type II bipolar illness
GM7262	3	EEMEM 20U	0308	21	F	WH		PROBAND	1013	1	Minor depression; one episode at age 20; father has type I bipolar illness; see GM7261 Lymphoid
GM7264	3	EEMEM 20U	0308	22	M	WH		PROBAND	1014	1	Type I bipolar illness; onset at age 20; 1 episode of depression; 1 episode of mania; hospitalized twice; on Lithium; father also has type I bipolar illness; see GM7263 Lymphoid
GM7266	2	EEMEM 20U	0308	17	F	WH		SIS	1014	2	See GM7265 Lymphoid; at risk; father and brother have type I bipolar illness
GM7248	2	EEMEM 20U	0308	17	F	WH		COUSIN	1021	2	At risk; one episode of minor depression at age 15; mother has type I bipolar illness; mother is dizygotic twin of mother of proband (GM7278 Lymphoid); see GM7247 Lymphoid
GM7260	2	EEMEM 20U	0308	24	M	WH		PROBAND	1022	1	Indeterminate number of episodes of hypomania; onset at age 9; no hospitalization; no medication; father has type II bipolar illness with major depression; mother has minor depression; see GM7259 Lymphoid
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM5926	2	EEMEM 20U	0432	50	M	WH		PROBAND	884	1	Type I bipolar illness; onset during teens; 12 hospitalizations for manic episodes & one for depression; illness has become chronic & less responsive to chemotherapy during recent years; see GM5927 Lymphoid

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DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM5928	2	EEMEM 20U	0432	48	F	WH		SPOUSE	884	2	Clinically unaffected; see GM5929 Lymphoid
GM5930	3	EEMEM 20U	0432	26	M	WH		SON	884	3	At risk; clinically normal; father and two sibs have type I bipolar illness; see GM5931A Lymphoid
GM5932	2	EEMEM 20U	0432	25	F	WH		AFF DAU	884	4	Type I bipolar illness; single episode at age 24; father & sib have type I bipolar illness; see GM5933 Lymphoid
GM5934	2	EEMEM 20U	0432	23	M	WH		AFF SON	884	5	Type I bipolar illness; drug & alcohol abuse; multiple admissions; chronic in recent years; possible schizoaffective; onset at age 21; father & sib have type I bipolar illness; see GM5935 Lymphoid
GM5936	2	EEMEM 20U	0432	18	M	WH		SON	884	7	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5937A Lymphoid
GM5938	2	EEMEM 20U	0432	16	M	WH		SON	884	8	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5939 Lymphoid
GM5940	2	EEMEM 20U	0432	14	F	WH		DAU	884	9	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5941 Lymphoid
GM5962A	8	EEMEM 20U	0432	86	M	WH		AFF FATHER	884	11	Probable type II bipolar illness; adult age of onset; semi-annual depressions lasting more than a week; see GM5963B Lymphoid
GM5960	2	EEMEM 20U	0432	82	F	WH		MOTHER	884	12	Some evidence of mild lability but no psychiatric diagnosis confirmed; one son has type I bipolar illness; see GM5961 Lymphoid
GM5904A	6	EEMEM 20U	0432	56	F	WH		SIS	884	13	Clinically normal; one brother, several nieces, and nephews have type I bipolar illness; see GM5903 Lymphoid
GM5974	2	EEMEM 20U	0432	55	F	WH		SIS	884	14	Clinically unaffected; one affected brother; one affected son (GM5976); spouse of GM5972; see GM5975A Lymphoid
GM5972	2	EEMEM 20U	0432	58	M	WH		AFF BRO-IN-L	884	15	Minor depressive disorder; early teens onset; has brief periods with some chronic minor depression lasting for months up to a year; spouse of GM5974; father of GM5976; see GM5973 Lymphoid

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				DISORDERS OF THE NERVOUS SYSTEM							
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM5970	2	EEMEM 20U	0432	61	F	WH		AFF IN-LAW	884	16	Minor depressive disorder; adult onset; brief intermittent depressive periods; paternal aunt of a type II bipolar individual; sister of GM5972; see GM5971 Lymphoid
GM5968	2	EEMEM 20U	0432	82	F	WH		IN-LAW	884	17	Clinically unaffected; two brothers have unipolar major depressive disorder; paternal grandmother of a type II bipolar individual; mother of GM5972; see GM5969 Lymphoid
GM5994	4	EEMEM 20U	0432	52	M	WH		AFF BRO	884	18	Personality disorder, unspecified, mixed with hypomanic features and anger dyscontrol; since adulthood has some role impairment; 2 children & a brother have type I bipolar illness; see GM5993 Lymphoid
GM5996	1	EEMEM 20U	0432	50	F	WH		AFF SIS-IN-L	884	19	Major depressive disorder; onset at age 30; 2 children & mother have type I bipolar illness; history of hospitalization; chemotherapy, & suicidal thoughts; daughter of GM6014 & GM6016; spouse of GM5994; see GM5995A Lymphoid
GM6014	2	EEMEM 20U	0432	77	M	WH		IN-LAW	884	20	Clinically unaffected; three affected children; father of GM5996; spouse of GM6016; see GM6013B Lymphoid
GM6016	2	EEMEM 20U	0432	74	F	WH		AFF IN-LAW	884	21	Type I bipolar illness; onset at age 24 yrs; recurrent episodes of depression & mania/hypomania; 3 affected children; mother of GM5996; spouse of GM6014; see GM6015A Lymphoid
GM6024	2	EEMEM 20U	0432	47	M	WH		IN-LAW	884	22	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6014 & GM6016; brother of GM5996; see GM6023 Lymphoid
GM6026	2	EEMEM 20U	0432	46	M	WH		AFF IN-LAW	884	23	Probable unipolar illness; onset at age 30 yrs; one major depressive episode characterized as situational; no psychosis; mother has type I bipolar illness; son of GM6014 & GM6016; brother of GM5996; see GM6025A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
					DISORDERS OF THE NERVOUS SYSTEM						
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM6028	3	EEMEM 20U	0432	41	M	WH		AFF IN-LAW	884	24	Type I bipolar illness; onset at age 27; intermittent periods of irritability and hypomania since single manic hospitalization; mother has type I bipolar illness; son of GM6014 & GM6016; brother of GM5996; see GM6027 Lymphoid
GM6018	2	EEMEM 20U	0432	39	M	WH		IN-LAW	884	25	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6014 & GM6016; brother of GM5996; see GM6017 Lymphoid
GM6030	2	EEMEM 20U	0432	37	F	WH		IN-LAW	884	26	Clinically unaffected; 3 affected sibs; mother nas type I bipolar illness; daughter of GM6014 & GM6016; sister of GM5996; see GM6029B Lymphoid
GM6012	2	EEMEM 20U	0432	33	M	WH		IN-LAW	884	27	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6014 & GM6016; brother of GM5996; see GM6011 Lymphoid
GM5897	3	EEMEM 20U	0432	44	M	WH		BRO	884	28	Clinically normal; brother has type I bipolar illness; father of GM5889, GM5891, GM5893, & GM5895; husband of GM5899; see GM5896 Lymphoid
GM5899	2	EEMEM 20U	0432	45	F	WH		SIS-IN-LAW	884	29	Clinically normal spouse of GM5897; mother of GM5889, GM5891, GM5893, & GM5895; see GM5898 Lymphoid
GM5906	1	EEMEM 20U	0432	33	M	WH		NEPHEW	884	30	Clinically normal; brother has type II bipolar illness; nephew and cousin of several type I bipolars; son of GM5972 & GM5974; see GM5905 Lymphoid
GM5984	2	EEMEM 20U	0432	27	F	WH		NIECE	884	31	Clinically unaffected; brother has type II bipolar illness; daughter of GM5972 & GM5974; see GM5983 Lymphoid
GM5976	2	EEMEM 20U	0432	26	M	WH		AFF NEPHEW	884	32	Type II bipolar illness; son of GM5972 & GM5974; see GM5977 Lymphoid
GM5986	2	EEMEM 20U	0432	20	F	WH		NIECE	884	33	Clinically unaffected; brother has type II bipolar illness; daughter of GM5972 & GM5974; see GM5985 Lymphoid
GM5978	1	EEMEM 20U	0432	17	F	WH		NIECE	884	34	At risk; clinically unaffected; brother has type II bipolar illness; daughter of GM5972 & GM5974; see GM5979 Lymphoid

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DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM5980	2	EEMEM 20U	0432	15	F	WH		NIECE	884	35	At risk; clinically unaffected; brother has type II bipolar illness; daughter of GM5972 & GM5974; see GM5981 Lymphoid
GM5988	1	EEMEM 20U	0432	30	F	WH		AFF NIECE	884	36	Type I bipolar illness; multiple admissions; similarly affected sister; dau. of GM5994 & GM5996; see GM5987A Lymphoid
GM5902	4	EEMEM 20U	0432	28	F	WH		NIECE	884	37	Clinically normal; two sibs with type I bipolar illness; daughter of GM5994 & GM5996; see GM5901 Lymphoid
GM5919	2	EEMEM 20U	0432	26	F	WH		NIECE	884	38	Clinically normal; two sibs have type I bipolar illness; daughter of GM5994 & GM5996; see GM5918 Lymphoid
GM5990	1	EEMEM 20U	0432	25	M	WH		NEPHEW	884	39	Clinically unaffected; two sibs have type I bipolar illness; son of GM5994 & GM5996; see GM5989 Lymphoid
GM5908	2	EEMEM 20U	0432	24	M	WH		NEPHEW	884	40	Clinically normal; two sibs have type I bipolar illness; son of GM5994 & GM5996; see GM5907 Lymphoid
GM5915	3	EEMEM 20U	0432	23	M	WH		NEPHEW	884	41	Clinically normal; two sibs have type I bipolar illness; son of GM5994 & GM5996; see GM5914 Lymphoid
GM6000	2	EEMEM 20U	0432	22	F	WH		AFF NIECE	884	42	Type I bipolar illness; onset at age 16 yrs; one manic episode and several major depressive episodes, each lasting from several weeks to a month; similarly affected sib; daughter of GM5994 & GM5996; see GM5999 Lymphoid
GM5992	2	EEMEM 20U	0432	20	F	WH		NIECE	884	43	Clinically unaffected; two sibs have type I bipolar illness; daughter of GM5994 & GM5996; see GM5991 Lymphoid
GM6002	2	EEMEM 20U	0432	19	M	WH		NEPHEW	884	44	Clinically unaffected; two sibs have type I bipolar illness; son of GM5994 & GM5996; see GM6001 Lymphoid
GM6004	1	EEMEM 20U	0432	16	F	WH		NIECE	884	46	Clinically unaffected; two sibs have type I bipolar illness; daughter of GM5994 & GM5996; see GM6003 Lymphoid
GM6006	2	EEMEM 20U	0432	12	F	WH		NIECE	884	47	At risk; clinically unaffected; two sibs have type I bipolar illness; daughter of GM5994 & GM5996; see GM6005 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)												
GM5895	2	EEMEM 20U	0432	22	M	WH			NEPHEW	884	48	Clinically normal with possible early signs of bipolar illness; second degree relative of several type I bipolar patients; son of GM5897 & GM5899; see GM5894 Lymphoid
GM5893	2	EEMEM 20U	0432	20	M	WH			NEPHEW	884	49	Clinically normal with brief periods of minor depression; second degree relative of several type I bipolar patients; son of GM5897 & GM5899; see GM5892 Lymphoid
GM5889	3	EEMEM 20U	0432	18	M	WH			NEPHEW	884	50	Clinically normal; second degree relative of several type I bipolar patients; son of GM5897 & GM5899; see GM5888 Lymphoid
GM5891	2	EEMEM 20U	0432	16	M	WH			NEPHEW	884	51	Clinically normal; second degree relative of several type I bipolar patients; son of GM5897 & GM5899; see GM5890 Lymphoid
Schizophrenia - 18150												
GM1792	3	EEMEM 20U	0137	26	M	WH	+-		PROBAND	119	2	See GM1793 Lymphoid; episodes of agitation, delusions of persecution, fear of assassination; father also affected
GM1835	3	EEMEM 20U	0137	27	F	WH	+-		AFF SIS	119	3	See GM1836 Lymphoid; hospitalized; drug abuse; father also affected; schizo-affective disorder
GM2497	3	EEMEM 20U	0137	23	M	WH	+-		PROBAND	176	1	See GM1488 Lymphoid; paralogical thinking, affective shielding, splitting of affect from content, suspiciousness; onset at age 15; hospitalized; positive family history
GM2503	2	EEMEM 20U	0137	27	F	WH	+-		AFF SIS	176	4	See GM1490 Lymphoid; anorexia nervosa since adolescence; more schizoid than depressed
GM2038	2	EEMEM 20U	0137	22	M	WH	+-		PROBAND	204	1	Skin biopsy; onset of symptoms at age 6; suicide; same patient as GM2039
GM2039	3	EEMEM 20U	0137	22	M	WH	+-		PROBAND	204	1	Liver biopsy fibroblast culture; same patient as GM2038; onset of symptoms at age 6; suicide
Spinocerebellar Ataxia												
GM4319	2	EEMEM 20U	0210	58	M	BL			PROBAND			Staggering wide based gait; type II muscle atrophy; ret. pig.; internuclear ophthalmoplegia; hyperactive DTR3+; bilat. ptosis; divergent strabismus; two sibs & maternal aunt affected; 46,XY/46,XY,del(13)(q14q22); 92%/8%

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				OPHTHALMOLOGIC DISORDERS							
Bardet-Biedl Syndrome - 20990											
GM5948	3	EEMEM 20U	0433	18	M	WH	--	PROBAND	891	1	Retinal degeneration; extinguished electro-retinograms; polydactyly; partial syndactyly; obesity at 5.5 years; similarly affected brother; see GM5952 Lymphoid
GM5949	3	EEMEM 20U	0433	50	F	WH	+-(O)	MOTHER	891	2	Clinically unaffected; two affected sons; see GM5951 Lymphoid
GM5950	2	EEMEM 20U	0433	19	M	WH	--	PROBAND	892	1	Retinal degeneration; extinguished electro-retinogram; obesity; polydactyly; mental retardation; see GM5953 Lymphoid
GM6081	3	EEMEM 20U	0433	46	F	WH	+-(O)	MOTHER	905	2	Clinically unaffected; 2 affected daughters; see GM6082 Lymphoid; proband is GM6080 Lymphoid
GM6196	6	EEMEM 20U	0433	34	M	WH	--	PROBAND	925	1	Pigmentary retinal degeneration; abnormal electro-retinograms; bilateral macular degeneration; posterior subcapsular lens opacities; mental retardation; nystagmus; polydactyly; see GM6195 Lymphoid
Choroideremia (Progressive Tapetochoroidal Dystrophy) - 30310											
GM6940	2	EEMEM 20U	0092	41	M	WH	Y-	PROBAND			Slowly progressive loss of vision with nightblindness; negative family history; striking atrophy of the pigment epithelium
Corneal Dystrophy, Macular Type - 21780											
*GM1125	3	EEMEM 20U	0092	41	F	WH	--	PROBAND			Corneal button fibroblast culture; mucopolysaccharidosis type unknown
Ectopia Lentis - 12960											
GM6812	2	EEMEM 20U	0092	28	F	WH	+-	PROBAND			Similarly affected son; progressive dislocation of the lens; glaucoma; bilateral surgical anophthalmos
Foveal Dystrophy, Progressive - 13655											
GM3785	3	EEMEM 20U	0351	49	M	WH	+-	PROBAND	534	1	20/70 vision, photophobia; positive family history; Deutean variant; relative scotomas in both eyes; see GM3784 Lymph.
Leber Optic Atrophy - 30890											
GM3858	4	EEMEM 20U	0351	33	M	WH	Y-	PROBAND	567	1	Onset in mid-teens; decreased bilateral visual acuity; similarly affected brother; see GM3857 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMD	FAMM	REMARKS
				OPHTHALMOLOGIC DISORDERS							
Aegurosis Congenita of Leber; Type I and II - 20400 & 20410											
GM3852	4	EEMEM 20U	0351	29MO	F	WH	--	PROBAND	564	1	Poor vision, nystagmus, 7 diopters hyperopic; similarly affected half sister but fathers were brothers; see GM3851 Lymphoid
Leopard Syndrome (Cardiomyopathic Lentiginosis) - 15110											
GM4107	2	EEMEM 20U	0351	14	M	WH	+-	PROBAND	646	1	Part of original family described by Gorlin; see GM4106 Lymphoid; mother similarly affected; growth retardation; sensory neural hearing loss; mild pulmonary stenosis; abnormal EKG
Lowe Oculocerebrorenal Syndrome - 30900											
*GM1676	2	EEMEM 20U	0107	10	M	WH	Y-	PROBAND			Positive family history; clinically affected
GM3265	2	EEMEM 20U	0107	17FW	M	WH	Y?	PROBAND	512	1	At risk (50%); 46,XY; a brother was affected; pedigree shows multiple affected individuals
Ophthalmoplegia, Pigment, Degen, of Retina & Cardiomyopathy - 16510											
GM6225	2	EEMEM 20U	0446	10	M	WH	+-	PROBAND	931	1	Kearns-Sayre syndrome; external ophthalmoplegia with retinal degeneration, ptosis, myocardial fibrosis with congestive heart failure, muscle weakness, diabetes, and absence of sweat; parents have normal ERGs & EKGs; see GM6224 Lym.
Pseudoxanthoma Elasticum - 17785 + 26480											
*GM4994	5	EEMEM 20U	0092	60	M	WH		PROBAND			Similarly affected sib; involvement includes eyes, skin, & gastrointestinal tract; neither parent showed signs of PXE; pedigree suggests recessive inheritance
Pterygium of Conjunctiva and Cornea - 17800											
GM4180	2	EEMEM 20U	0351	34	M	WH	+-	PROBAND	681	1	Unilateral; 4 affected generations; onset at age 12; see GM4181 Lymphoid
Retinoblastoma, Hereditary - 18020											
GM1262	4	EEMEM 20U	0198	7WK	F	WH	+-	PROBAND			Bilateral; conjunctiva fibroblast culture
*GM1879A	7	EEMEM 20U	0155	11	F	WH	+-	PROBAND	118	1	Skin fibroblast culture; bilateral; also has osteosarcoma; 46,XX; mother has unilateral retinoblastoma
*GM1880	3	EEMEM 20U	0155	37	F	WH	+-	AFF MOTHER	118	2	Unilateral retinoblastoma; benign right para-ovarian cyst at 24; multiple leiomyomas of uterus at 29; 46,XX in leukocytes; skin fibroblast culture; moderate hypersensitivity to cell killing by ionizing radiation and bleomycin

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				OPHTHALMOLOGIC DISORDERS								
Retinoblastoma; Hereditary - 18020												
GM3313	3	EEMEM 20U	0155	40	M	WH	++		FATHER	118	3	Skin fibroblast culture; 46,XY/45,X; (76%/24%); clinically unaffected
*GM1408	4	EEMEM 20U	0198	8MO	F	WH	+-		PROBAND	185	1	Bilateral; left eye conjunctiva fibroblast culture
*GM1123	3	EEMEM 20U	0094	9MO	F	BL	+-		PROBAND	189	1	Bilateral; early onset; multifocal; identical twin of GM1131; no other family members affected; 46,XX; conjunctiva fibroblast culture
*GM1131	3	EEMEM 20U	0094	9MO	F	BL	+-		AFF ID TWIN	189	2	Bilateral; early onset; multifocal; identical twin of GM1123; no other family members affected; 46,XX; conjunctiva fibroblast culture
GM6418	3	EEMEM 20U	0092	30	M	WH	+-		PROBAND	960	1	Persian; bilateral, familial retinoblastoma; skin fibroblast culture; see GM6417 Lymphoid; 46,XY; similarly affected son
GM6419	2	EEMEM 20U	0092	18MO	M	WH	+-		AFF SON	960	2	Persian; bilateral stage IV retinoblastoma; diagnosed at age 8 mos.; skin fibroblast culture; 46,XY; similarly affected father
*GM1232		R1640 20I	0198	30MO	F	WH	+-		PROBAND	993	1	Y79; tumor tissue culture, grows in suspension; passage 3 at IHR
Retinoblastoma; Sporadic												
*GM2718	7	EEMEM 15U	0244	7	F		+?	A	PROBAND			46,XX,del(13)(pter>q12.3::q21.2>qter)
GM2971	14	EEMEM 20U	0240	5MO	F	WH	+?	A	PROBAND			46,X,t(X;13)(Xqter>Xp22::13q12>13qter; 13pter>13q12::Xp22>Xpter); balanced; abnormal X is late replicating
GM4250	4	EEMEM 20U	0373	22MO	F	OR	+?		PROBAND			Japanese; bilateral; 46,XX,t(13;X)(q12;p22) in leukocytes; translocated X preferentially inactivated; erythrocyte esterase D Type 1-1 in proband, sister and parents
GM0914	3	EEMEM 20U	0198	2	M	WH	+?		PROBAND	117	1	Unilateral; conjunctiva fibroblast culture; 46,XY,9qh+; same patient as GM0913
*GM1142A	8	EEMEM 20U	0094	27MO	F	WH	+?	A	PROBAND	187	1	Bilateral; 46,XX,del(13)(pter>q14.1::q22.1>qter); see GM1484 Lymphoid and GM3022 SV40 Transformed Fibroblast; 1.6 times more sensitive than normal cells to cell killing by ionizing radiation; nor sensitivity to DNA alkylating agents

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
					OPHTHALMOLOGIC DISORDERS							
Retinoblastoma, Sporadic												
GM5877	8	EEMEM 20U	0047	2	F		+?	A	PROBAND	1025	1	46,XX,del(13)(pter>q14.1::q21.2>qter) in lymphocytes and fibroblasts; bilateral; sporadic; negative family history; normal growth and development except for delayed speech; see GM7312 Lymphoid
Retinitis Pigmentosa - 18010, 26800, 31260												
GM3824	2	EEMEM 20U	0351	50	M	WH			PROBAND	554	1	Mild optic atrophy with severe pigmentary retinopathy; ring scotoma; extinguished ERG; pedigree shows dominant inheritance; see GM3823A Lymphoid
GM3826	4	EEMEM 20U	0351	22	M	WH			AFF SON	554	2	Mild optic atrophy with moderate pigmentary retinopathy; pedigree shows dominant inheritance; see GM3825 Lymphoid
GM3831	4	EEMEM 20U	0351	56	F	WH			PROBAND	558	1	Fundus shows pigmentary degenerative type changes; nonrecordable ERG; ring scotomas both eyes; dominant inheritance; see GM3832 Lymphoid
GM3835	4	EEMEM 20U	0351	32	F	WH			AFF DAU	558	2	Mild abnormal pigmentation; subnormal ERG; dominant inheritance; see GM3836 Lymphoid
GM3833	3	EEMEM 20U	0351	32	F	WH			PROBAND	559	1	Night blindness; subnormal ERG; pedigree shows dominant inheritance; see GM3834 Lymphoid
GM3837	2	EEMEM 20U	0351	12	F	WH			AFF DAU	559	2	Poor night vision; moderately subnormal ERG; dominant inheritance; see GM3838 Lymphoid
GM3893	3	EEMEM 20U	0351	36	M	WH			PROBAND	573	1	Similarly affected father and brother; probably dominant inheritance; see GM3894 Lymphoid
GM3906	3	EEMEM 20U	0351	5	M	WH			PROBAND	578	1	General tapetoretinal degeneration; pedigree suggests X-linked inheritance; see GM3907 Lymphoid
GM3910	3	EEMEM 20U	0351	41	F	WH			AFF MOTHER	578	2	High myopia; constricted visual field; pedigree suggests X-linked inheritance; extinguished ERG; see GM3911 Lymphoid
GM7236	2	EEMEM 20U	0466	34	F	WH			PROBAND	775	1	Pedigree suggests recessive inheritance; see GM7235 Lymphoid; onset at age 3; severely myopic; pigmentary degeneration; loss of visual acuity; typical fundus findings; constricted visual fields; similarly affected brother

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				OPHTHALMOLOGIC DISORDERS							
Retinitis Pigmentosa - 18010, 26800, 31260											
GM7232	4	EEMEM 20U	0466	74	M	WH		FATHER	775	2	See GM7231 Lymphoid; clinically unaffected; two affected children; pedigree suggests recessive inheritance
GM7234	3	EEMEM 20U	0466	68	F	WH		MOTHER	775	3	See GM7233 Lymphoid; clinically unaffected; two affected children; pedigree suggests recessive inheritance
GM7369	2	EEMEM 20U	0466	45	M	WH		BRO	775	4	See GM7368 Lymphoid; clinically unaffected; two affected sibs; pedigree suggests recessive inheritance
GM7483	3	EEMEM 20U	0466	47	M	WH		AFF BRO	775	5	Bilateral; onset at age 5; loss of visual acuity; constricted visual fields; similarly affected sister; pedigree suggests recessive inheritance; see GM7482 Lymphoid
Usher Syndrome - Retinitis Pigmentosa + Congenital Deafness - 27690											
GM3854	6	EEMEM 20U	0351	47	M	WH	--	PROBAND	565	1	German; mildly affected; slow progression; brother is similarly affected; see GM3853 Lymphoid; 46,XY
GM3889	4	EEMEM 20U	0351	22	M	WH	--	PROBAND	572	1	Progressive tapetoretinal degeneration; similarly affected sib; see GM3890A Lymphoid; 46,XY
GM3891	3	EEMEM 20U	0351	16	M	WH	--	AFF BRO	572	2	Progressive tapetoretinal degeneration; see GM3892A Lymphoid
				OTHER DISORDERS OF KNOWN BIOCHEMISTRY							
Acatalasia (Acatalasemia; Catalase Deficiency) - 20020											
GM1931	8	EEMEM 20U	0169	73	M	WH	+-(D)	PARENT			Swiss; father of an affected child; proband not in Repository
Analbuminemia - 20530											
*GM6248	7	EEMEM 20I	0091	12	F	AI	--	PROBAND			Trace amounts of apparently normal serum albumin; non-edematous; atopic dermatitis; mild bronchial asthma; mild seizure disorder; hyperlipoproteinemia with corneal arcus; consanguineous parents; 62% of cells show 46,XX,inv(11)
Fragile Site 10q25 - 13662											
*GM5425A	14	EEMEM 20U	0418	28	F	WH	A	PROBAND	855	1	See GM5523A Lymphoid; fra(10)(q25) observed in 12% of cells at 917; short stature, late menarche, irregular menses, small uterus, small ovaries, aortic insufficiency, myopia, and cataracts

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Porphyria: Hereditary Coproporphyria - 12130												
GM0962	3	EEMEM 20U	0112	59	F	WH	+-		PROBAND	641	1	Clinically affected; 46,XX; see GM2302 Lymphoid
Porphyria: Acute Intermittent Porphyria - 17600												
GM1647	3	EEMEM 20U	0112	44	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase
GM7077	2	EEMEM 20U	0126	32	M	BL	+-		PROBAND			Paralysis; normal erythrocyte porphobilinogen deaminase activity and immunologically cross reacting material; urine contains large amounts of PBG
*GM0933	3	EEMEM 20U	0112	15MO	F	WH	+-	A	DAU	81	1	Latent carrier; deficient URO synthetase
*GM0932	3	EEMEM 20U	0112	4	M	WH	+-	A	SON	81	2	Latent carrier; deficient URO synthetase
*GM0931	3	EEMEM 20U	0112	32	F	WH	+-	A	PROBAND	81	3	Active AIP; deficient URO synthetase; positive family history
*GM0939	3	EEMEM 20U	0112	32	F	WH	+-	A	PROBAND	82	1	See GM2135 Lymphoid; active AIP; deficient URO synthetase; positive family history
*GM0940	2	EEMEM 20U	0112	61	F	WH	++	A	MOTHER	82	2	See GM2134 Lymphoid; clinically normal; no URO synthetase deficiency
*GM0941	3	EEMEM 20U	0112	63	M	WH	+-	A	FATHER	82	3	Latent carrier; see GM2133 Lymphoid; deficient URO synthetase
GM1623	2	EEMEM 20U	0112	16	M	WH	+-	A	AFF BRO	83	1	Deficient uroporphyrinogen-I synthetase
GM1622	3	EEMEM 20U	0112	19	F	WH	+-	A	PROBAND	83	2	Deficient uroporphyrinogen-I synthetase; similarly affected brother and mother
*GM1621	2	EEMEM 20U	0112	39	F	WH	+-	A	AFF MOTHER	83	3	Deficient uroporphyrinogen-I synthetase; two affected children
GM1625	3	EEMEM 20U	0112	3	M	WH	+-	A	AFF SON	84	1	Deficient uroporphyrinogen-I synthetase
GM1624	3	EEMEM 20U	0112	22	F	WH	+-	A	PROBAND	84	2	Deficient uroporphyrinogen-I synthetase; positive family history
Porphyria: Cutanea Tarda - 17610												
GM0961	3	EEMEM 20U	0112	70	M	WH	+-		PROBAND			Clinically affected
GM0977	5	EEMEM 20U	0112	81	F	WH	+-		PROBAND			Clinically affected
GM1041	6	EEMEM 20U	0112	69	F	WH	+-		PROBAND			Clinically affected

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Porphyria: Cutanea Tarda - 17010												
GM1179	3	EEMEM 20U	0112	36	M	WH	+-		PROBAND			Clinically affected
GM1482	4	EEMEM 20U	0112	58	M	WH	+-		PROBAND			Clinically affected
Protoporphyria, Erythropoietic; Heme Synthetase Deficiency - 17700												
*GM5008A	22	EEMEM 20U	0396	26	F	WH	+-	B	PROBAND			Photosensitivity since infancy; deficient heme synthetase activity; normal delta-aminolevulinic acid synthetase activity; similarly affected sister
*GM5005A	19	EEMEM 20U	0396	15	F	WH	+-	B	PROBAND	807	1	Photosensitivity since infancy; deficient heme synthetase activity; normal delta-aminolevulinic acid synthetase activity; positive family history
*GM5007A	15	EEMEM 20U	0396	40	M	WH	+-	B	AFF FATHER	807	3	Photosensitivity since infancy; deficient heme synthetase activity; normal delta-aminolevulinic acid synthetase activity; two affected children
Porphyria, Congenital Erythropoietic (Gunther Disease) - 26370												
GM1082A	5	EEMEM 20U	0112	21	M	WH	--		PROBAND			Diagnosis based upon characteristic porphyrins in erythrocytes, urine, & feces
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
AGR Triad (Aniridia, Genitourinary Abnormalities; Mental Retardation)- 19407												
GM3809	2	EEMEM 20U	0320	2	F	WH			PROBAND			Aniridia, genitourinary abnormalities, & mental retardation; XX phenotype - 46,XY karyotype (X-linked familial testicular feminization); pedigree contains several females with no menses
GM6938B	3	EEMEM 20U	0148	3WK	M	WH		A	PROBAND	1074	1	Micropenis; crypterchidism; bilateral aniridia; optic nerve atrophy; genital skin fibroblast culture; 46,XY,del(11)(pter>p13::p14>qter); see GM7736 Lymphoid
Antitrypsin Deficiency of Plasma (Alpha-1-Antitrypsin) - 10740												
GM2522	2	EEMEM 20U	0107	3MO	M	WH	--		PROBAND			Serum Pi type is ZZ; both parents are MZ
Basal Cell Nevus Syndrome (Odontogenic Keratocysts & Skeletal Anom.) - 10940												
GM3300	2	EEMEM 20U	0107	11	F	WH	+-		PROBAND			Jaw cysts; cleft lip and palate; mental retardation; multiple nevi
GM1657	3	EEMEM 20U	0104	53	M	WH	+-		PROBAND	88	1	Normal skin; 46,XY; 14% of cells show random chromosome loss; see GM1656 Lymphoid; same patient as GM1658

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Basal Cell Nevus Syndrome (Odontogenic Keratocysts & Skeletal Anom.) - 10940												
GM1658	3	EEMEM 20U	0104	53	M	WH	+-		PROBAND	88	1	Basal cell carcinoma; see GM1656 Lymphoid; same patient as GM1657; 46,XY; 18% of cells show random translocations
GM1725	3	EEMEM 20U	0104	58	F	WH	+-		PROBAND	89	1	See GM1726 Lymphoid; clinically affected
*GM2098	3	EEMEM 20U	0104	31	M	WH	+-		PROBAND	90	1	See GM2099 Lymphoid; clinically affected; fibroblasts are hypersensitive to cell killing by ionizing radiation
GM2138	4	EEMEM 20I	0104		F	WH	+-		PROBAND	91	1	See GM2139 Lymphoid; clinically affected
GM1577A	6	EEMEM 20U	0104	39	M	WH	+-		PROBAND	372	1	See GM1578 Lymphoid; clinically affected
*GM1575	2	EEMEM 20U	0104	31	F	WH	+-		PROBAND	373	1	See GM1576 Lymphoid; clinically affected; fibroblasts are hypersensitive to cell killing by ionizing radiation
*GM1552	2	EEMEM 20U	0104	27	M	WH	+-		PROBAND	637	1	See GM1553 Lymphoid; clinically affected; fibroblasts are hypersensitive to cell killing by ionizing radiation
Beckwith-Wiedemann Syndrome (EMG Syndrome) - 13065												
*GM0097	3	EEMEM 20U	0107	1	F	WH	+-	A	PROBAND			De novo translocation; 46,X,t(X;1)(Xpter>Xq26::1q12>1qter;1pter>1q12::Xq26>Xqter); balanced; normal X is late replicating
GM0359	3	EEMEM 20U	0107	8	M	WH	+-		PROBAND	351	1	Negative family history; clinically affected
Chediak-Higashi Syndrome (Natural Killer Lymphocyte Defect) - 21450												
GM2075	3	EEMEM 20U	0126	18MO	F	WH	--		PROBAND			Photophobia; no nystagmus; cytoplasmic inclusions in WBC; partial oculocutaneous albinism
Coffin-Lowry Syndrome - 30360												
*GM2365	4	EEMEM 20U	0213	14	M	WH	Y-	A	PROBAND			Mental retardation; short stature; unusual facies; tapering fingers; positive family history; 46,XY,inv(2)(pter>p11.2::q13>p11.2::q13>qter); balanced
GM3318	2	EEMEM 20U	0107	8	M	WH	Y-		PROBAND	510	1	See GM3317 Lymphoid; mental retardation; coarse facies; positive family history; 46,XY
Cholestasis with Peripheral Pulmonary Stenosis - Alagille Syndrome - 11845												
GM5759	3	EEMEM 20U	0107	3MO	F	WH	+-		PROBAND			Hepatoductular hypoplasia; paucity of intrahepatic bile ducts; myocardial fibrous scarring; peripheral pulmonary stenosis; characteristic facies; jaundiced at birth

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMS	FAMN	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
*GM0342A	6	EEMEM 20U	0107	14	M	WH	--	PROBAND			Advanced with pulmonary involvement at the time of diagnosis at age 6; expired at age 14; 46,XY
*GM0770A	7	EEMEM 20U	0060	19	M	WH	--	PROBAND			Classic pulmonary and pancreatic involvement; no diabetes; one spontaneous pneumothorax
*GM0997	8	EEMEM 20U	0059	10	M	WH	--	PROBAND			Diagnosed at age 7 mos; negative family history for CF; sweat chloride: 120 meq/L; on enzyme replacements; expired at age 11
*GM0998	8	EEMEM 20U	0059	4	M	WH	--	PROBAND			Confirmed by dexamethasone resistance; sweat chloride: 126 meq/L; on enzyme therapy
*GM0999	14	EEMEM 20U	0059	13	F	WH	--	PROBAND			Diagnosed at age 3; one affected and six normal sibs; sweat chloride: 144 meq/L; symptoms are GI and pulmonary
*GM1348	3	EEMEM 20U	0107	18	F	WH	--	PROBAND			Severe; first diagnosed at age 7; expired at age 18; 46,XX
GM1958	4	EEMEM 20U	0178	13	M	WH	--	PROBAND			Diagnosed at age 3 mo.; very little pulmonary involvement; a deceased sib had C.F.
GM2801	5	EEMEM 20U	0060	15	M	WH	--	PROBAND			Biopsy from nasal polyp; pancreatic insufficiency; elevated sweat electrolytes
GM2828		EEMEM 20U	0060	26	F	WH	--	PROBAND			Advanced pulmonary involvement; pneumothorax; no pancreatic insufficiency; a sib died of C.F. in early infancy with meconium ileus; passage 4 at IMR
*GM1012A	12	EEMEM 20U	0005	8	M	WH	--	PROBAND	92	1	Elevated sweat chloride; similarly affected sib
*GM1011	10	EEMEM 10U	0005	7	M	WH	--	AFF BRO	92	2	Elevated sweat chloride
*GM1014A	14	EEMEM 20U	0005	13	M	WH	--	PROBAND	93	1	Elevated sweat chloride; similarly affected sib
*GM1013	9	EEMEM 20U	0005	11	M	WH	--	AFF BRO	93	2	Elevated sweat chloride
GM1708	12	EEMEM 20U	0005	33	M	WH	+-(O)	FATHER	93	3	Formerly GM1010; two affected sons

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
*GM1957	6	EEMEM 20U	0178	11	M	WH	--	PROBAND	94	1	Diagnosed at age 3 mo.; pancreatic insufficiency; similarly affected sib
*GM1959	4	EEMEM 20U	0178	10	M	WH	--	AFF BRO	94	2	Also suffers from asthma
GM3465	3	EEMEM 20U	0107	4	M	WH	--	PROBAND	233	1	Pericardium biopsy; has two affected sibs; first diagnosed by sweat test at age 10 weeks; pulmonary and hepatic cirrhosis; expired at age 4; 46,XY; same patient as GM3466
GM3466	2	EEMEM 20U	0107	4	M	WH	--	PROBAND	233	1	Skin biopsy; has two affected sibs; first diagnosed by sweat test at age 10 weeks; pulmonary and hepatic cirrhosis; expired at age 4; same patient as GM3465
*GM1707	10	EEMEM 20U	0005	9	F	WH	--	PROBAND	253	1	Confirmed by dexamethasone resistance; formerly GM1008
GM2827A	5	EEMEM 20U	0060	23	F	WH	--	PROBAND	336	1	Expired at age 25; pancreatic insufficiency and diabetes mellitus; elevated sweat chloride
GM2826A	13	EEMEM 20U	0060	51	F	WH	+-(0)	MOTHER	336	2	Normal sweat electrolytes
GM2803	5	EEMEM 20U	0060	27	M	WH	+?	BRO	336	3	Unaffected; possible heterozygote; normal sweat electrolytes; sister died at age 25 of C.F.
*GM0768	3	EEMEM 20U	0060	13	M	WH	--	PROBAND	404	1	Mild; no pancreatic insufficiency; first diagnosed at age 4 mos; a sib died from C.F. at age 10; see GM0897 Lymphoid; 46, XY; 4% of cells show random chromosome loss
*GM0851A	12	R1640 20U	0025	22	M	WH	--	PROBAND	426	1	See GM1531 Lymphoid; clinically affected
GM4320	2	EEMEM 20U	0060	17	M	WH	--	PROBAND	704	1	Advanced pulmonary disease; pancreatic insufficiency; pneumothorax; no diabetes; elevated sweat electrolytes
GM4322	2	EEMEM 20U	0060	17	F	WH	--	PROBAND	705	1	Advanced pulmonary disease; pancreatic insufficiency; no diabetes; malnutrition; elevated sweat electrolytes; a sib died of C.F. at age 7
GM4323	4	EEMEM 20U	0060	45	F	WH	+-(0)	MOTHER	705	2	Normal sweat electrolytes; two affected children

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM4337	3	EEMEM 20U	0060	50	M	WH	+-(O)	FATHER	705	3	Normal sweat electrolytes; two affected children
GM4339	3	EEMEM 20U	0060	24	M	WH	--	PROBAND	706	1	Advanced chronic pulmonary disease; no pancreatic involvement; elevated sweat electrolytes; a sib died at age 4 with meconium ileus
GM4338	3	EEMEM 20U	0060	47	F	WH	+-(O)	MOTHER	706	2	Normal sweat electrolytes; two affected children
GM4331	2	EEMEM 20U	0283	20	M	BL	--	PROBAND	711	1	Classic C.F. with pancreatic insufficiency; elevated sweat electrolytes; see GM4330 Lymphoid
GM4346	2	EEMEM 20U	0283	25	M	WH	--	PROBAND	712	1	Classic C.F. with pancreatic insufficiency and diabetes mellitus; elevated sweat electrolytes; see GM4345 Lymphoid
GM4539	3	EEMEM 20U	0060	18	F	WH	--	AFF SIS	765	1	Chronic cough; abnormal sweat electrolytes; absence of stool proteolytic enzymes; peribronchial thickening and sinus opacification; staph. aureus in the sputum; acute onset Insulin requiring diabetes
GM4541	3	EEMEM 20U	0060	10	M	WH	--	PROBAND	765	2	Abnormal sweat electrolytes; minimal cough & congestion; mild pulmonary symptomatology; little or no stool proteolytic enzyme activity; see GM4540 Lymphoid
GM4543	3	EEMEM 20U	0060	39	F	WH	+-(O)	MOTHER	765	3	Two affected children
GM4547	3	EEMEM 20U	0060	43	M	WH	+-(O)	FATHER	765	4	Two affected children
GM4545	3	EEMEM 20U	0060	20	M	WH	+?	BRO	765	5	Normal sweat electrolytes; possible heterozygote; two affected siblings
Diabetes Mellitus of the Pima Indian - 22210											
GM2557	3	EEMEM 20U	0215	29	F	AI		PROBAND			Both parents and 3 grandparents are diabetic; onset at age 28
GM2574	3	EEMEM 20U	0215	23	M	AI		PROBAND	230	1	Pima/Papago American Indian; onset at age 18; on Insulin; see GM2575 Lymphoid
GM2565	2	EEMEM 20U	0215	28	M	AI		PROBAND	231	1	Onset at age 27 yrs; not on Insulin; see GM2566 Lymphoid

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus of the Pima Indian - 22210											
*GM2416	3	EEMEM 20U	0215	42	F	AI		PROBAND	383	1	See GM2417 Lymphoid; mother and two sibs also diabetic
*GM2440	2	EEMEM 20U	0215	23	M	AI		PROBAND	384	1	Both parents are also diabetic
GM2442	4	EEMEM 20U	0215	36	F	AI		PROBAND	385	1	Both parents are also diabetic
GM2556	2	EEMEM 20U	0215	27	F	AI		PROBAND	386	1	Onset at age 26; obese; not on Insulin; mother also diabetic
Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585											
*GM1237	3	EEMEM 20U	0034	29	M	WH		PROBAND	95	1	MODY; onset at age 11; see GM1247A Lymphoid; 6 affected and 4 unaffected sibs
*GM1219	5	EEMEM 20U	0034	46	M	WH		AFF BRO	95	2	MODY; onset at age 29; see GM1246 Lymphoid
*GM1122	3	EEMEM 20U	0034	33	F	WH		AFF SIS	95	3	MODY; onset at age 16; see GM1240 Lymphoid
*GM1430	3	EEMEM 20U	0034	35	M	WH		AFF BRO	95	4	MODY; onset at age 27; see GM1243 Lymphoid
*GM1955	3	EEMEM 20U	0034	43	M	WH		AFF BRO	95	5	MODY; onset at age 25; see GM1956 Lymphoid
*GM1497	2	EEMEM 20U	0034		F	WH		AFF SIS	95	6	MODY; onset at age 30; see GM1498 Lymphoid
*GM1435	5	EEMEM 20U	0034	22	M	WH		AFF NEPH	95	7	MODY; onset at age 14; see GM1242 Lymphoid; son of GM1497
*GM1496	3	EEMEM 20U	0034	20	M	WH		AFF NEPH	95	8	MODY; onset at age 12; see GM1244 Lymphoid; son of GM1497
*GM1409	3	EEMEM 20U	0034	15	F	WH		AFF NIECE	95	9	Maternal cousin of GM1435 & GM1496; only juvenile diabetic in family; onset at age 13; on Insulin; see GM1410 Lymphoid
*GM1837	4	EEMEM 20U	0034	22	F	WH		AFF COU	95	10	MODY type II; onset at age 18; paternal 1st cousin once removed of the 6 sibs; see GM1838 Lymphoid
*GM4420	7	EEMEM 20U	0034	47	F	WH		SIS	95	11	Non-diabetic; 7 affected sibs
*GM4421	6	EEMEM 20U	0034	29	F	WH		SIS	95	12	Non-diabetic; 7 affected sibs
*GM4422	5	EEMEM 20U	0034	45	F	WH		SIS	95	13	Non-diabetic; 7 affected sibs

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY							
Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585											
GM1486	3	EEMEM 20U	0034	37	M	WH		PROBAND	291	1	Third generation of MOD; see GM1241A Lymphoid
Diabetes Mellitus: Juvenile Onset - 22210											
*GM0609	3	EEMEM 20U	0100	15	F	WH		PROBAND			On Insulin; clinically affected
*GM2637	4	R1640 20U	0095	17	M	WH		PROBAND	236	1	Onset at age 2; on Insulin; similarly affected sib; see GM2676 Lymphoid
*GM2638	4	R1640 20U	0095	10	F	WH		AFF SIS	236	2	Onset at age 3; on Insulin; see GM2793A Lymphoid
*GM2639	6	R1640 20U	0095	19	M	WH		BRO	236	3	Non-diabetic; two affected sibs; see GM2675 Lymphoid
*GM2640	4	R1640 20U	0095	41	M	WH		FATHER	236	4	Non-diabetic; see GM2741B Lymphoid; both parents had maturity onset diabetes; two affected children
*GM2641	5	R1640 10U	0095	41	F	WH		MOTHER	236	5	Non-diabetic; two affected children; see GM3355 Lymphoid
*GM2712	8	R1640 10U	0095	17	M	WH		PROBAND	243	1	Onset at age 14; on Insulin; two affected & three unaffected sibs; see GM2713A Lymphoid
*GM2704	9	R1640 10U	0095	45	M	WH		FATHER	243	2	Non-diabetic; three affected children; see GM2705 Lymphoid
*GM2706	11	R1640 10U	0095	45	F	WH		MOTHER	243	3	Non-diabetic; three affected children; see GM2707 Lymphoid
*GM2708	9	R1640 10U	0095	16	M	WH		BRO	243	4	Non-diabetic; three affected sibs; see GM2709 Lymphoid
*GM2710	12	R1640 10U	0095	10	M	WH		BRO	243	5	Non-diabetic; three affected sibs; see GM2711 Lymphoid
*GM2869	7	R1640 20U	0260	25	F	WH		SIS	243	6	Non-diabetic; three affected sibs; see GM2728 Lymphoid
*GM2868	7	R1640 20U	0260	24	M	WH		AFF BRO	243	7	Onset at age 22
*GM2956A	10	EEMEM 10U	0260	23	M	WH		AFF BRO	243	8	Onset at age 11; see GM3027 Lymphoid
*GM2209	4	EEMEM 10U	0095	31	F	WH		SIS	307	1	Non-diabetic; HLA type A1,B8; mother has Insulin-controlled and father has diet-controlled diabetes

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
*GM2635		R1640 20U	0095	33	F	WH		PROBAND	307	2	Onset at age 10; on Insulin; HLA type A1,A2/B8,Bw15; passage 2 at IMR
*GM3358	5	R1640 10U	0260	15	M	WH		PROBAND	606	1	Onset at age 14; on Insulin; positive family history; see GM3235 Lymphoid
*GM3359	5	R1640 10U	0260	13	M	WH		BRO	606	2	Non-diabetic; father and brother are diabetics; see GM3239 Lymphoid
*GM3360	5	R1640 10U	0260	43	M	WH		AFF FATHER	606	3	Onset at age 14; on Insulin; see GM3236 Lymphoid
*GM3361	6	R1640 10U	0260	17	M	WH		BRO	606	4	Non-diabetic; father and brother are diabetics; see GM3237 Lymphoid
*GM3362	6	R1640 10U	0260	41	F	WH		MOTHER	606	5	Non-diabetic; spouse and son are diabetics; see GM3238B Lymphoid
*GM1872	3	EEMEM 20U	0095	12	F	WH		PROBAND	620	1	Onset at age 10; on Insulin; three affected sibs; see GM2762 Lymphoid
*GM1873	3	EEMEM 20U	0095	15	F	WH		AFF SIS	620	2	Onset at age 13; on Insulin; see GM2763 Lymphoid
*GM1909	3	EEMEM 20U	0095	19	M	WH		AFF BRO	620	3	Onset at age 4; on Insulin; hypertension; short stature; see GM2765 Lymphoid
*GM1910	3	EEMEM 20U	0095	21	F	WH		SIS	620	4	Non-diabetic; four affected sibs; see GM3354A Lymphoid
*GM1911	3	EEMEM 20U	0095	20	M	WH		AFF BRO	620	5	Onset at age 4; on Insulin; see GM2764 Lymphoid
*GM1875	4	EEMEM 20U	0095	17	M	WH		BRO	620	6	Non-diabetic; four affected sibs; see GM2866A Lymphoid
*GM1874	4	EEMEM 20U	0095	22	F	WH		SIS	620	7	Non-diabetic; mentally retarded; four affected sibs; see GM2766 Lymphoid
*GM1876	4	EEMEM 20U	0095	18	M	WH		BRO	620	8	Non-diabetic; four affected sibs; see GM2812A Lymphoid
*GM1878	4	EEMEM 20U	0095	41	F	WH		MOTHER	620	9	Non-diabetic; four affected children; see GM2714 Lymphoid
*GM1877	4	EEMEM 20U	0095	50	M	WH		FATHER	620	10	Non-diabetic; four affected children; see GM3356A Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAMB	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Diabetes Mellitus: Juvenile Onset - 22210												
*GM2613	8	R1640 10U	0095	24	F	WH			PROBAND	621	1	Onset at age 6; similarly affected brother; see GM2677 Lymphoid
*GM2614	8	R1640 10U	0095	27	M	WH			AFF BRO	621	2	Onset at age 18 mo.; on Insulin; see GM2678 Lymphoid
*GM2615	8	R1640 10U	0095	29	M	WH			BRO	621	3	Non-diabetic; two affected sibs; see GM2679 Lymphoid
*GM2616	8	R1640 10U	0095	63	M	WH			FATHER	621	4	Non-diabetic; see GM2680 Lymphoid; two affected children
*GM2617	7	R1640 10U	0095	60	F	WH			MOTHER	621	5	Non-diabetic; see GM2785A Lymphoid; two affected children
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230												
*GM1610	3	EEMEM 20U	0095	13	F	WH			PROBAND	96	1	Onset at age 11; optic atrophy; on Insulin; two affected sibs; see GM1795 Lymphoid
*GM1611	4	EEMEM 20U	0095	15	F	WH			AFF SIS	96	3	Onset at age 13; optic atrophy; on Insulin
*GM1701	4	EEMEM 20U	0095	42	M	WH			FATHER	96	4	Non-diabetic; three affected children; see GM1797 Lymphoid
DiGeorge Syndrome - 10840												
*GM2944A	4	EEMEM 20U	0107	2WK	M	WH	+-	A	PROBAND			46,XY,-22,+der(20),t(20;22)(20pter>20q11.2::22q11.2>22qter); unbalanced; clinically affected
GM3479	3	EEMEM 20U	0107	2MO	M	WH	+-		PROBAND			46,XY; absent thymus, TE fistula, esophageal atresia; bilateral cleft lip and palate; rocker bottom feet
*GM3577	2	EEMEM 20U	0107	8MO	M	WH	+-	A	PROBAND			45,XY,-22,t(3;22)(3pter>3q29::22q11>22qter); unbalanced; absent thymus & parathyroid glands; cleft lip & palate; congenital heart disease
*GM5401	3	EEMEM 20U	0417	3MO	M	WH	+-	A	PROBAND			Hispanic; partial DiGeorge syndrome; truncus arteriosis, type I; T-cell dysfunction; micrognathia and high arched palate; positive family history; 45,XY,-4,-22,+der(4),t(4;22)(4pter>4q35::22q11>22qter)mat; unbalanced
GM5876	5	EEMEM 20U	0148	6MO	M	WH	+-		PROBAND			46,XY in lymphocytes and fibroblasts; complex congenital heart disease including interrupted aortic arch and VSD; neonatal hypocalcemia; seizures; absent thymus; intermittent lymphopenia; small ears; possible facial palsy

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
DiGeorge Syndrome - 18840											
GM7215	3	EEMEM 20U	0148	3DA	F	WH	+-	PROBAND			Interrupted aortic arch; ASD; VSD; absent thymus (radiographic); 46,XX in lymphocytes and fibroblasts
Dubowitz Syndrome - 22337											
GM2850	2	EEMEM 20U	0269	2	M	AI	--	PROBAND			46,XY; peculiar facies; short stature; sparse hair; mental retardation; negative family history
Dyskeratosis Congenita; Zinsser-Cole-Engman Syndrome - 30500											
*GM1774	2	EEMEM 20U	0107	7	M	WH	Y-	PROBAND	99	1	See GM1775 Lymphoid; maternal grandfather was also affected; leukoplakia tongue; dysplastic fingernails & toenails; increased chromosome instability in BrdU treated fibroblasts; hypersensitive to mitomycin-C induced cell killing
GM1786	2	EEMEM 20U	0107	30	F	WH	+-(D)	MOTHER	99	2	Affected father and son
GM1787	2	EEMEM 20U	0107	78	F	WH	+?	MAT GR GRMR	99	3	Possible carrier; affected son & great-grandson
Eosinophilic Fascitis											
GM2870	2	EEMEM 20U	0293	64	M	WH		PROBAND			May synthesize abnormal collagen or mucopolysaccharide
Epidermodysplasia Verruciformis - 22640											
GM2984	11	EEMEM 20U	0248		M	WH	--	PROBAND			French; warts; squamous cell carcinoma of skin; negative family history
GM2972	10	EEMEM 20U	0011		M	WH	--	PROBAND	511	1	Erupted skin; higher than normal UV induced unscheduled DNA synthesis; same patient as GM2973
GM2973	8	EEMEM 20U	0011		M	WH	--	PROBAND	511	1	Uninvolved skin; same patient as GM2972
Gout - 13890											
GM0432	3	EEMEM 20U	0120	32	M	WH	+-	PROBAND			Severe gouty arthritis
Possible Hair-Brain Syndrome (Amish Brittle Hair Syndrome) - 23405											
GM6331	1	EEMEM 20U	0319	16	M	WH	--	PROBAND	954	1	Moroccan; consanguineous parents; growth and mental deficiency; brittle hair; absent puberty; microcephaly; areas of alopecia; hypotonia; two similarly affected sibs; normal urinary organic acids and plasma biotinidase activity
GM6332	1	EEMEM 20U	0319	4	F	WH	--	AFF SIS	954	2	Moroccan; consanguineous parents; growth and mental deficiency; brittle hair; microcephaly; areas of alopecia; hypotonia; two similarly affected sibs; normal urinary organic acids and plasma biotinidase activity

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
							DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY				
Possible Hair-Brain Syndrome (Amish Brittle Hair Syndrome) - 23405											
GM6333	1	EEMEM 20U	0319	9MO	F	WH	--	AFF SIS	954	3	Moroccan; consanguineous parents; failure to thrive; psychomotor retardation; brittle hair; microcephaly; areas of alopecia; hypotonia; two similarly affected sibs; normal urinary organic acids and plasma biotinidase activity
Jeune Syndrome: Newborn Asphyxiating Thoracic Dystrophy - 20850											
GM3521	2	EEMEM 20U	0074	2	F	WH	--	PROBAND			46,XX; thoracic-pelvic-phalangeal dystrophy
Lipoatrophic Diabetes (Seip Syndrome; Berardinelli Syndrome) - 26970											
*GM0990B	14	EEMEM 20U	0055	15	M	WH	--	PROBAND	797	1	Original diagnosis was atypical progeria; patient has hypertriglyceridemia and abnormal glucose tolerance test; line KH; 46,XY; 25% of fibroblasts show chromosome or chromatid breakage
*GM1828	4	EEMEM 20U	0178	18	M	WH	--	PROBAND	797	1	Original diagnosis was atypical progeria; line KH; same patient as GM0990B
Meckel Syndrome - 24900											
GM7817	4	EEMEM 20I	0518	24FW	F	BL	--	PROBAND			Abortus skin biopsy; encephalocele; polydactyly; polycystic kidneys; contractures; 46,XX in peripheral blood lymphocytes
GM3206	2	EEMEM 20U	0107	NB	F	WH	--	PROBAND	640	1	Skin biopsy; occipital encephalocele; polycystic kidneys; microcephaly; cultures from muscle and pericardium also available; 46,XX,9qh+
Macrocephaly, Multiple Lipomas, and Hemangiomata - 15348											
GM4910	8	EEMEM 20U	0270	4	F	WH	+-	PROBAND			No hemangiomas; low-normal IQ; delayed expressive language skills; three affected generations
Neurofibromatosis (von Recklinghausen Disease) - 16220											
*GM0622	3	EEMEM 20U	0095	8	M		+-	PROBAND			Clinically affected
*GM1633	3	EEMEM 20U	0104	61	M	WH	+-	PROBAND	113	1	Atypical; see GM1634 Lymphoid
GM1639	3	EEMEM 20U	0104	19	F	BL	+-	PROBAND	114	1	Normal skin; other fibroblasts also available; see GM1641 Lymphoid
GM1858	2	EEMEM 20U	0104	41	M	WH	+-	PROBAND	115	1	Neurofibroma biopsy-fibroblast culture; see GM1861 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Neurofibromatosis (von Recklinghausen Disease) - 16220											
GM1859	3	EEMEM 20U	0104	41	M	WH	+-	PROBAND	115	1	Cafe au lait spot biopsy-fibroblast culture; same patient as GM1858; see GM1861 Lymphoid
GM1860A	6	EEMEM 20U	0104	41	M	WH	+-	PROBAND	115	1	Normal skin; see GM1861 Lymphoid; same patient as GM1858
Papular Mucinosis (genetic or acquired)											
GM0948	3	EEMEM 20U	0124	60	M	WH		PROBAND	116	1	Non-lesioned gingiva; same patient as GM0950 & GM0951
GM0950	2	EEMEM 20U	0124	60	M	WH		PROBAND	116	1	Non-lesioned leg skin; same patient as GM0948
GM0951	3	EEMEM 20U	0124	60	M	WH		PROBAND	116	1	Lesioned leg skin; same patient as GM0948
Polyposis, Intestinal: Type I (Familial Polyposis of the Colon) - 17510											
*GM2355	2	EEMEM 20U	0212	13	F	WH	+-	PROBAND	479	1	Aff. father, grandmother, greatgrandfather, & greatgreatgrandmother; numerous sm. polyps present in the cecum, rectum, & sigmoid; cult. is derived from a skin (arm) biopsy; 46,XX,22p+; sl. hypersensitivity to UV induced cell killing
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
GM2694A	11	EEMEM 20U	0225	40	F	WH	+-	PROBAND	525	1	Tubulovillus adenoma; polyps; 46,XX; 27% of fibroblasts are polyploid
GM3900	3	EEMEM 20U	0358	15	F	BL	+-	PROBAND	582	1	Abdominal tumor biopsy; Gardner syndrome; similarly affected father; 14% of cells are polyploid at P8; 46,XX,inv(9)(pter>p11::q13>p11::q13>qter); balanced; same patient as GM3901
GM3901	3	EEMEM 20U	0358	15	F	BL	+-	PROBAND	582	1	Skin biopsy; Gardner syndrome; similarly affected father; 11% of cells are polyploid at P5; 46,XX,inv(9)(pter>p11::q13>p11::q13>qter); balanced; same patient as GM3900
*GM3223	2	EEMEM 20U	0225	25	M	WH	+-	AFF SON	603	1	See GM3224 Lymphoid; normal sensitivity to MNNG produced loss of colony-forming ability; 46,XY; 8% of fibroblasts are 46,XY,t(1;10)(q32p15); unbalanced; 7% are polyploid
*GM3314	2	EEMEM 20U	0225	48	F	WH	+-	PROBAND	603	2	English; adenocarcinoma of colon; see GM3315A Lymphoid; hypersensitivity to MNNG produced loss of colony-forming ability; 46,XX/46,XX,t(5;15)(5pter>5q22::15q26>15qter;15pter>15q26::5q22>5qter); 40%/60%; 14% are polyploid at P5

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530												
GM3940	3	EEMEM 20U	0225	45	M	WH	+?		BRO	603	3	At risk; see GM3941 Lymphoid; 46,XY; 12% of cells show random chromosome loss; 2% of cells show chromosome aberrations
GM3944	5	EEMEM 20U	0225	38	F	WH	+-		AFF SIS	603	4	Cysts, polyps, ileoproctostomy; see GM3945 Lymphoid; hypersensitivity to MNNG produced loss of colony-forming ability; 46,XX; 8% of cells are polyploid
GM3942	4	EEMEM 20U	0225	17	F	WH	+?		NIECE	603	5	At risk; daughter of GM3944; see GM3943 Lymphoid; 46,XX; 10% of cells show random chromosome loss; 4% of cells show chromosome aberrations
GM3946	2	EEMEM 20U	0225	22	F	WH	+-		AFF DAU	603	6	See GM3947 Lymphoid; hypersensitivity to MNNG produced loss of colony-forming ability; 46,XX; 10% of cells show random chromosome loss; 11% are polyploid
*GM3948	3	EEMEM 20U	0225	30	F	WH	+-		AFF DAU	603	7	Cysts, polyps, and desmoid tumor; see GM3949 Lymphoid; hypersensitivity to MNNG produced loss of colony-forming ability; 46,XX; 6% of cells show chromosome aberrations
GM3950	4	EEMEM 20U	0225	12	F	WH	+?		DAU	603	8	At risk; 46,XX/46,XX,t(12;13)(12pter>12q11::13q34>13qter;13pter>13q34::12q11>12qter); balanced; 10%/90%; see GM3951A Lymphoid; 3 affected sibs and mother
GM3952	3	EEMEM 20U	0225	20	F	WH	+?		NIECE	603	9	At risk; unaffected parents; father is GM3940; see GM3953 Lymphoid; 46,XX; 2% of cells show chromosome breaks
GM3954	3	EEMEM 20U	0225	24	M	WH	+-		AFF SON	603	10	Polyps and cysts; see GM3955 Lymphoid; normal sensitivity to MNNG produced loss of colony-forming ability; 46,XY
GM3956	3	EEMEM 20U	0225	27	F	WH	++		DAU-IN-LAW	603	11	Clinically normal; 46,XX; 4% of cells show chromosome aberrations; see GM3957 Lymphoid
*GM6855	7	EEMEM 20U	0468	19	M		+-		PROBAND	967	1	Fibroma fibroblast culture; dental abnormalities; multiple polyposis; multiple osteomas; multiple fibromas; multiple epidermoid cysts; desmoids; see GM7500 Lymphoid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
*GM6856	15	EEMEM 20U	0468	14	M		+-	AFF COU	967	2	Multiple polyposis; multiple osteomas; multiple fibromas; multiple epidermoid cysts; dental abnormalities
*GM6888A	7	EEMEM 20U	0468	16	M		+-	AFF COU	967	4	Desmoid fibroblast culture; multiple polyposis; multiple osteomas; multiple fibromas; multiple epidermoid cysts; dental abnormalities; brother of GM6856; 46,XY; 22% of cells show chromatid breaks; 10% show random loss
*GM6964	6	EEMEM 20U	0468	20	F	WH	+-	AFF SIS	967	5	Polyps, desmoids, fibromas, epidermoid cysts, osteomas, dental abnormalities, & adenomas; skin fibro cult; 46,XX; no chromo #2 abnormality seen at P9; 12% of cells show random chromo loss/gain; 10% of cells are polyploid; see GM7501 Lymph
*GM6965	8	EEMEM 20U	0468	55	F	WH	+-	AFF MOTHER	967	6	Polyps, desmoids, fibromas, epidermoid cysts, osteomas, dental abnormalities, & adenomas; affected father, brother, and children; skin fibroblast culture; 46, XX; 6% of cells show breaks, 12% show random loss; see GM7499 Lymphoid
Progeria (Hutchinson - Gilford Syndrome) - 17667											
GM0989B	15	EEMEM 20U	0055	20	M		+-	PROBAND			Atypical progeria with unspecified cachectic dwarfism
*GM1177B		EEMEM 20U	0049	9	M		+-	PROBAND			From Canadian Repository; 46,XY; passage 15 at IMR; clinically affected
GM1178	18	EEMEM 20U	0049	34	M		+-	PROBAND			From Canadian Repository; slow growing culture; patient died at age 34; small eyes; micrognathia; nystagmus; large tongue; cardiac murmur
*GM1972A	12	EEMEM 20U	0091	14	F	WH	+-	PROBAND			Classical progeria; 46,XX in lymphocytes and fibroblasts; 6% of cells show chromosome aberrations; same pt. as AG1972A
Reticulosis, Familial Histiocytic - 26770											
GM7125	1	EEMEM 20U	0074	2	M	WH	--	PROBAND			CNS degeneration; recurrent fever; hepatomegaly; anemia; neutropenia; thrombocytopenia; liver and retro-orbital tissue biopsy demonstrated histiocytic infiltration

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Sideroblastic Anemia												
*GM4516	3	EEMEM 20U	0074	5	F	WH			PROBAND	736	1	Pancytopenia, pancreatic insufficiency, dystonia, and neurological symptoms; see GM4515 Lymphoid
Smith-Lemli-Opitz Syndrome (RSH Syndrome) - 27040												
GM3044	3	EEMEM 20U	0148	2WK	A	WH	--	A	PROBAND			45,XY,t(13;14)(13qter>cen>14qter); unbalanced in fibroblasts; clinically affected
GM5788	2	EEMEM 20U	0204	17MO	F	WH	--		PROBAND			46,XX in leukocytes; failure to thrive, developmental delay, hypotonia, microcephaly, micrognathia, simian creases, & partial syndactyly; negative family history; see GM5789 Lymphoid
Telangiectasia, Hereditary Hemorrhagic (Rendu-Osler-Weber Syndrome) -18730												
GM3418	2	EEMEM 20U	0210	52	M	WH	+-		AFF PAT GRFR	211	1	Gastrointestinal bleeding; mucous telangiectasias; 4 affected generations
GM3419	2	EEMEM 20U	0210	7	F	WH	+-		AFF DAU	211	2	Mucocutaneous telangiectasias; 4 affected generations
GM3015	3	EEMEM 20U	0210	28	M	WH	+-		PROBAND	211	3	Rendu-Osler-Weber Disease; seizures; cutaneous, ocular, and gastric telangiectasias; father, daughter, and grandfather were also affected
Tetralogy of Fallot - 18750												
GM3126	2	EEMEM 20U	0148	4	F	BL	+-	A	PROBAND			Microcephaly; mental retardation; absent uterus, cervix, and upper vagina; ureteral reflux; otitis media; cyanotic congenital heart disease; 46,XX,del inv(1)(pter>p13::q21>p13::q21>q42::), unbalanced
GM7151	7	EEMEM 20I	0489	2WK	F	WH	+-	A	PROBAND			Multiple congenital defects including cleft palate, coloboma, tetralogy of Fallot, and abnormal genitalia and extremities; 46,X,t(X;17)(Xpter>Xq11.2::17q11.2>17qter::17pter>17q11.2::Xq11.2>Xqter); balanced
Tuberous Sclerosis - 19110												
*GM1635	3	EEMEM 20U	0104	17	M	BL	+-		PROBAND	124	1	Shagreen patch; see GM1636 Lymphoid; same patient as GM1643
GM1643	3	EEMEM 20U	0104	17	M	BL	+-		PROBAND	124	1	Normal skin; see GM1636 Lymphoid; same patient as GM1635

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Tuberous Sclerosis - 19110											
GM1637	2	EEMEM 20U	0104	20	F	BL	+-	PROBAND	125	1	Ashleaf spot; same patient as GM1644; see GM1638 Lymphoid
*GM1644	2	EEMEM 20U	0104	20	F	BL	+-	PROBAND	125	1	Normal skin; see GM1638 Lymphoid; same patient as GM1637
GM2332	2	EEMEM 20U	0210	23	M	WH	+-	PROBAND	216	1	Normal skin; seizures; mental retardation; shagreen patches; adenoma sebaceum; same patient as GM2333
GM2333	4	EEMEM 20U	0210	23	M	WH	+-	PROBAND	216	1	Subungual fibroma fibroblast culture; same patient as GM2332
Wilms Tumor (Nephroblastoma) - 19407											
GM3118	2	EEMEM 20U	0320		F		+- A	PROBAND			Bilateral; bilateral aniridia; new intrarenal mass; skin fibroblast culture; 46,XX,del(11)(p13p11)/46,XX,del(11)(p13 p11),t(7;22)(p15;p12); 40%/60%
GM3808	2	EEMEM 20U	0320	9	M	WH	+- A	PROBAND			AGR triad; seizures; Wilms tumor; 46,XY, del(11)(pter>p13::p11>qter)
Wiskott-Aldrich Syndrome - 30100											
*GM1598	2	EEMEM 20U	0107	4	M	WH	Y-	PROBAND			Clinically affected; no family history of the disease
Zellweger Syndrome (Cerebrohepatorenal Syndrome) - 21410											
GM0228	3	EEMEM 20U	0107	2MO	M	WH	-- A	PROBAND			Clinically affected; increased C26:0 and C26:1 long-chain fatty acids and C26:C22 fatty acid ratio in fibroblasts
*GM4340	2	EEMEM 20U	0006	2MO	F	WH	-- A	PROBAND			Spanish; severe failure to thrive; typical facies; generalized amino-aciduria; epiphyseal stippling in the knees; child is product of a father-daughter mating; eye findings include zonular cataracts, abs. ERG & abnor. VER; elevated C26:C22
GM6093	6	EEMEM 20U	0437	16MO	M	WH	--	PROBAND			Hypotonia, developmental delay, mental retardation, optic hypoplasia, macrocephaly, and cirrhosis; increased serum iron level; absent hepatic peroxisomes
GM6094	15	EEMEM 20U	0437	30MO	F	WH	--	PROBAND			Failure to thrive; abnormal facies, hypotonia, hepatomegaly, cirrhosis, small calcification at the patella, developmental delay, and mental retardation; absent hepatic peroxisomes; slow growing culture

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Zellweger Syndrome (Cerebrohepatorenal Syndrome) - 21410											
GM6231	2	EEMEM 20U	0447	1MO	F	WH	--	PROBAND			Consanguineous parents; similarly affected sib; muscle hypotonia, craniofacial dysmorphia, ventricular septal defect, glossoptosis, cataracts, hepatomegaly with jaundice and elevated SGOT & SGPT, and elevated CSF protein; 46,XX
GM6256		EEMEM 20U	0452	7MO	F	WH	--	PROBAND			Delayed develop; hepatomegaly; rotary nystagmus; large ant fontanelle; protuberant eyes; questionable retinitis pigmentosa; passage 3 at IMR; 46,XX,inv(9)(pter>p11::q21>p11::q21>qter)pat; balanced; increas C26:C22 fatty acid ratio
X CHROMOSOME MARKERS											
Agammaglobulinemia (Bruton Type) - 30030											
*GM0362	3	EEMEM 20U	0125	21	M	WH	Y-	PROBAND			Clinically affected; 46,XY in leukocytes and fibroblasts; normal RBC G6PD level; Xq(a) antigen positive
GM3654	7	EEMEM 20U	0352	26	M	WH	Y-	PROBAND			Russian-Italian; infantile, Bruton-type agammaglobulinemia; three affected generations
Glucose-6-Phosphate Dehydrogenase Deficiency & Variants - 30590											
*GM0120	9	EEMEM 20U	0041	30	F	BL	A	PROBAND			G6PD-A,PGK-1/G6PD-B,PGK-2; the majority of cells in culture express the genes for G6PD-B & PGK-2; 46,XX; 18% of cells show random chromosome loss/gain at P11
GM0324	5	EEMEM 20U	0125	22	M	WH	A	PROBAND			Sardinian; homozygous for G6PD (med. type); Xg(a) antigen present; 47,XY, t(X;7)(Xpter>Xq24::7q32>7qter;7pter>7q32::Xq24>Xqter)
GM0325	4	EEMEM 20U	0125	30	M	WH	A	PROBAND			Sardinian; heterozygous for G6PD deficiency (med. type), Xg(a) antigen present; 47,XXY
*GM0412	3	EEMEM 20U	0009	19	M	WH		PROBAND			G6PD deficiency; Panama type; 46,XY
*GM0738	7	EEMEM 20U	0041		M	WH		PROBAND			Hektoen variant of G6PD; 46,XY; 18% of cells are polyploid at passage 9
GM1165	2	EEMEM 20U	0125	37	F	WH		PROBAND			46,XX; double heterozygote G6PD(B) Xg(a)-/G6PD(med) Xg(a)+

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			X CHROMOSOME MARKERS									
Glucose-6-Phosphate Dehydrogenase Deficiency 6 Variants - 30590												
*GM1166	3	EEMEM 20U	0125	39	F	WH			PROBAND			46,XX; double homozygote, G6PD(B) Xg(a)-/ G6PD(B) Xg(a)-; 0.75 units G6PD in fibroblasts
*GM3165	2	EEMEM 20U	0117	8	M	WH			PROBAND			Hemolysis; RBC G6PD deficient; Greek; 0.15 units G6PD in fibroblasts
*GM3171	2	EEMEM 20U	0117	16	M	WH		A	PROBAND			Hemolysis; RBC G6PD deficient; Greek; 0.10 units G6PD in fibroblasts
GM3172	3	EEMEM 20U	0117	39	F	WH			PROBAND			Favism; hemolysis; RBC G6PD deficient; Greek
*GM3173	4	EEMEM 20U	0117	43	M	WH			PROBAND			Absent red cell G6PD; Greek; 0.14 units G6PD in fibroblasts
*GM3174	3	EEMEM 20U	0117	40	F	WH			PROBAND			Absent red cell G6PD; Greek; 0.13 units G6PD in fibroblasts
GM3175	2	EEMEM 20U	0117	36	M	WH			PROBAND			Hemolysis; RBC G6PD deficient; 0.11 units G6PD in fibroblasts; Greek
GM1150	4	EEMEM 20U	0125	23	M	WH			PROBAND	157	1	46,XY; normal RBC G6PD(B) level, Xg(a) antigen absent; Sardinian
GM1152	4	EEMEM 20U	0125	49	M	WH			FATHER	157	2	46,XY; RBC deficient for G6PD(med type), Xg(a) antigen positive; Sardinian
GM1163	4	EEMEM 20U	0125	15	M	WH			BRO	157	3	46,XY; deficient RBC G6PD, Xg(a) antigen positive; Sardinian
GM1871	3	EEMEM 20U	0125	47	F	WH			MOTHER	157	4	46,XX; double heterozygote; G6PD(B) Xg(a)+/G6PD(med type) Xg(a)-; Sardinian
GM1151	3	EEMEM 20U	0125	12	M	WH			PROBAND	158	1	46,XY; RBC deficient for G6PD(med type), Xg(a) antigen absent; Sardinian
GM1154	4	EEMEM 20U	0125	37	F	WH			MOTHER	158	2	46,XX; double heterozygote; G6PD(med type) Xg(a)-/ G6PD(B) Xg(a)+; Sardinian
*GM1153	4	EEMEM 20U	0125	42	M	WH			FATHER	158	3	46,XY; normal RBC G6PD level, Xg(a) antigen positive; Sardinian; 0.53 units G6PD in fibroblasts; slow growing culture
GM1188	3	EEMEM 20U	0125		M	WH			PROBAND	159	1	46,XY,inv(9); G6PD(Seattle), Xg(a) antigen positive; Sardinian; 24% of cells have marker chromosome 11q+ at P5

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS												
Glucose-6-Phosphate Dehydrogenase Deficiency & Variants - 30590												
GM1157	5	EEMEM 20U	0125	42	F	WH			MOTHER	159	2	46,XX; double heterozygote G6PD(Seattle) Xg(a)+/ G6PD(B) Xg(a)-; Sardinian
GM1193	4	EEMEM 20U	0125	50	F	WH			PROBAND	160	1	46,XX; RBC are heterozygous G6PD(B)/ G6PD(med) but are lacking enzyme activity; Sardinian
GM1192A	7	EEMEM 20U	0125	70	F	WH			MOTHER	160	2	46,XX; RBC are heterozygous G6PD(B)/G6PD (med) but have normal levels of enzyme activity; Sardinian
GM2334	4	EEMEM 20U	0125		F	WH			MOTHER	217	2	46,XX; heterozygote; G6PD(B)/G6PD(med), Xg(a) antigen positive; son is GM2335 Fibroblast
GM2336	3	EEMEM 20U	0125		M	WH			FATHER	217	3	46,XY; G6PD(B), Xg(a) antigen positive; son is GM2335 Fibroblast
GM1886	3	EEMEM 20U	0125	11	M	WH			PROBAND	290	1	46,XY; G6PD(med), Xg(a) antigen absent
*GM1887	3	EEMEM 20U	0125	7	M	WH			BRO	290	2	46,XY; G6PD(B), Xg(a) antigen absent; 0.76 units G6PD in fibroblasts
GM1888	2	EEMEM 20U	0125	14	M	WH			BRO	290	3	46,XY; G6PD(B), Xg(a) antigen absent
*GM1869	2	EEMEM 20U	0125	15	M	WH			PROBAND	504	1	46,XY; normal RBC G6PD(B) level, Xg(a) antigen absent; 0.69 units G6PD in fibroblasts
GM1870	2	EEMEM 20U	0125	14	M	WH			MAT 1ST COU	504	2	46,XY; RBC deficient for G6PD(med), Xg(a) antigen absent
Ichthyosis: X-Linked; Steroid Sulfatase Deficiency - 30810												
*GM3034	2	EEMEM 20U	0125	10	M	WH	Y-	A	PROBAND			Sterol sulfatase deficiency; G6PD(med), Xg(a) antigen absent
GM3273	3	EEMEM 20U	0125	34	M	WH	Y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) negative; Sterol sulfatase deficient
GM3276	3	EEMEM 20U	0125	13	M	WH	Y-	B	PROBAND			Sardinian; G6PD deficient, Xg(a) positive; Sterol sulfatase deficient
*GM3277	2	EEMEM 20U	0125	15	M	WH	Y-	A	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient
GM3278	3	EEMEM 20U	0125	26	M	WH	Y-	B	PROBAND			Sardinian; G6PD normal, Xg(a) positive; Sterol sulfatase deficient

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS												
Ichthyosis: X-Linked; Steroid Sulfatase Deficiency - 30810												
GM3281	3	EEMEM 20U	0125	48	F	WH	+?	B	PROBAND			Sardinian; G6PD(med)+ Xg(a)-/ G6PD(B)+ Xg(a)+; normal Sterol sulfatase
GM3283	3	EEMEM 20U	0125	37	F	WH	+?	B	PROBAND			Sardinian; G6PD(B)+ Xg(a)-/G6PD(med)+ Xg(a)+; Sterol sulfatase normal
GM3289	3	EEMEM 20U	0125	27	M	WH	Y-	B	PROBAND			Sardinian; G6PD normal+ Xg(a) positive; Sterol sulfatase deficient
GM3291	3	EEMEM 20U	0125	60	M	WH	Y-	B	PROBAND			Sardinian; G6PD normal+ Xg(a) positive; Sterol sulfatase deficient
GM3030	2	EEMEM 20U	0125	9	M	WH	Y-	A	PROBAND	434	1	Sterol sulfatase deficiency; normal RBC G6PD; Xg(a) antigen positive
GM3031	5	EEMEM 20U	0125	47	F	WH	+?	A	MOTHER	434	2	G6PD(B)/G6PD(med); normal Sterol sulfatase
GM3033	5	EEMEM 20U	0125	66	F	WH	+-(O)	A	PARENT	435	2	G6PD(B)/G6PD(med); normal Sterol sulfatase; mother of 3 affected sons; proband not in Repository
GM3035	4	EEMEM 20U	0125	20	M	WH	Y-	A	PROBAND	436	1	Sterol sulfatase deficiency; G6PD(med); Xg(a) antigen absent
GM3280	4	EEMEM 20U	0125	46	M	WH	Y?	B	PROBAND	815	1	Sardinian; G6PD normal; Xg(a) positive; sterol sulfatase normal; possible variant; possibly not X-linked ichthyosis
Mental Retardation, X-linked, Nonspecific - 30953												
GM1228	3	EEMEM 20U	0066	12	M	WH	Y-		PROBAND	642	1	Positive family history; formerly GM0642; 46,XY; fra(X) was not identified in 100 cells examined
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955												
GM5282A	3	EEMEM 20U	0224	21FW	M	BL	Y-	B	PROBAND			Skin fibroblast culture; oldest half-brother is mentally retarded and expresses fra(X); another half-brother is mentally retarded; mother does not express the fra(X); 6% of fibroblasts show 46,fra(X),Y
GM7730A	4	EEMEM 20U	0512	20FW	M	WH	Y-	B	PROBAND			Placental fibroblast culture; two affected maternal uncles have fra(X)(q27); 6% of cultured amniotic fluid cells and 26% of placental fibroblasts showed the fra(X)

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS												
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955												
*GM4024A	7	EEMEM 20U	0061	29	M	BL	Y-	A	PROBAND	588	1	Seizures; macro-orchidism; mental retardation; 52% 46,fra(X)(q27),Y in leukocytes; 4% in fibroblasts using FudR; G6PD type A in blood; see GM4025B Lymphoid
*GM4026	2	EEMEM 20U	0061	35	M	WH	Y-	A	PROBAND	589	1	Macro-orchidism; mental retardation; 46,fra(X)(q27),Y in leukocytes; 2% in fibroblasts; positive family history
*GM4926	4	EEMEM 20U	0061	42	M	WH	Y-		PROBAND	780	1	Mental retardation & macro-orchidism; 46,fra(X)(q2800),Y in leukocytes; similarly affected brother and cousins
GM5131	6	EEMEM 20U	0148	3	M	WH	Y-	A	PROBAND	819	1	Similarly affected brother & uncle; 46,fra(X)(q27),Y in leukocytes; 8% of fibroblasts show fra(X) with FudR
GM5185	6	EEMEM 20U	0390	26	M	WH	Y-		AFF MAT UNC	819	3	46,fra(X),Y present in 30-50% of leukocytes
GM5848A	5	EEMEM 20U	0427	4	M	WH	Y-	A	PROBAND	872	1	Moderate mental retardation; large ears and long face with prognathic appearance; undefined connective tissue dysplasia; 50% of lymphocytes and 12% of fibroblasts show 46,fra(X)(q27),Y with FudR
GM5847	6	EEMEM 20U	0427	36	F	WH	+-(0)	A	MOTHER	872	2	Clinically normal but dull; mild connective tissue dysplasia; 40% of lymphocytes and 0% to 4% of fibroblasts show 46,fra(X)(q27),X; one affected son
GM7071	2	EEMEM 20U	0061	22FW	M	WH	Y-		PROBAND	987	1	Kidney biopsy; mother is an obligate carrier for fra(X); 9/50 cord blood lymphocytes showed fra(X); mother is GM6968 Lymphoid; same pt. as GM7072; negative for fra(X) in kidney tissue
GM7072	1	EEMEM 20U	0061	22FW	M	WH	Y-		PROBAND	987	1	Lung biopsy; mother is an obligate carrier for fra(X); 9/50 cord blood lymphocytes showed fra(X); mother is GM6968 Lymphoid; same pt. as GM7071; negative for fra(X) in lung tissue
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890												
*GM0367A	5	EEMEM 20U	0107	17	M	WH	--		PROBAND			Fibroblast culture (lung biopsy); a deceased sister had A.T.; more resistant to neocarzinostatin inhibition of DNA

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SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890											
GM0647A	8	EEMEM 20U	0133	17	M	WH	--	PROBAND			Neurologic disability & dramatic telangiectasia; elevated serum alpha-fetoprotein; slow growing culture
*GM0648B	18	EEMEM 20U	0133	15	M	WH	--	PROBAND			Neurologic disability and dramatic telangiectasia; random chromosome abnormalities in 60% of cells karyotyped; 46,XY
*GM1588	13	EEMEM 20U	0077		M	WH	--	PROBAND			46,XY; clinically affected
GM1829	10	EEMEM 20U	0133	7	M	WH	--	PROBAND			Clinically affected
GM1841	11	EEMEM 20U	0133	13	M		--	PROBAND			Clinically affected
*GM2052	3	EEMEM 20U	0077	15	F	WH	--	PROBAND			46,XX; Jewish-Moroccan; fibroblasts have decreased post x-ray irradiation colony-forming ability; AT19IJE-F; decreased post-neocarzinostatin exposure colony-forming ability
GM7481	12	EEMEM 20U	0056	5	M	WH	--	PROBAND			AT1PX; cerebellar ataxia; questionable telangiectases; chronic respiratory infection; low IgA; developed Burkitt lymphoma; expired at age 5; 46,XY, with numerous rearrangements and/or deletions & increased breakage
GM2797	4	EEMEM 20U	0109	40	F	WH	+-(0)	PARENT	13	1	Amish; mother of an affected child; proband not in Repository
*GM1937	7	EEMEM 10U	0077	24	F	WH	--	PROBAND	126	2	Jewish-Moroccan; AT17IJE-F; decreased post-neocarzinostatin exposure colony-forming ability; similarly affected sister
*GM2531	6	EEMEM 20U	0077	5	M	WH	--	PROBAND	228	1	Iranian; see GM2782 Lymphoid; similarly affected sib; AT20IJE-F; decreased post-neocarzinostatin exposure colony-forming ability
*GM2530	7	EEMEM 20U	0077	8	M	WH	--	AFF BRO	228	2	Iranian; see GM2783A Lymphoid; AT21IJE-F; decreased post-neocarzinostatin exposure colony-forming ability
*GM1740A	21	EEMEM 20U	0077		M	WH	--	PROBAND	419	1	Moroccan; a deceased sister had A.T.; same patient as GM1939
*GM3488	3	EEMEM 20U	0226	41	M	WH	+-(0)	FATHER	516	1	Clinically normal; ATH15Be; two affected children; see GM3187 Lymphoid; normal sensitivity to cell killing by X-irradiation

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
							SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE					
							Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890					
*GM3489	2	EEMEM 20U	0226	37	F	WH	+-(O)		MOTHER	516	2	Clinically normal; ATH16Be: two affected children; see GM3188 Lymphoid; normal sensitivity to cell killing by X-irradiation
GM3487B	6	EEMEM 20U	0226	8	M	WH	--		PROBAND	516	3	AT4Be; neurological abnormalities; similarly affected sib; see GM3189 Lymphoid; increased sensitivity to cell killing by X-irradiation
GM3490	3	EEMEM 20U	0226	16	F	WH	+?		SIS	516	4	See GM3324 Lymphoid; possible heterozygote; clinically unaffected
GM3492	2	EEMEM 20U	0226	7	M	WH	+?		BRO	516	5	See GM3323 Lymphoid; possible heterozygote; clinically unaffected
GM3491	2	EEMEM 20U	0226	15	F	WH	+?		SIS	516	6	See GM3325 Lymphoid; possible heterozygote; clinically unaffected
*GM3395A	7	EEMEM 20U	0226	13	M	BL	--		PROBAND	605	1	See GM3332 Lymphoid; a deceased sister had A.T.; increased sensitivity to cell killing by X-irradiation
*GM3397	2	EEMEM 20U	0226	43	M	BL	+-(O)		FATHER	605	2	See GM3382 Lymphoid; clinically normal; two affected children; normal sensitivity to cell killing by X-irradiation
*GM3396	2	EEMEM 20U	0226	42	F	BL	+-(O)		MOTHER	605	3	See GM3334 Lymphoid; clinically normal; two affected children; normal sensitivity to cell killing by X-irradiation
GM3400	2	EEMEM 20U	0226	11	F	BL	+?		SIS	605	4	Normal; possible heterozygote; see GM3333 Lymphoid
GM3399	3	EEMEM 20U	0226	20	F	BL	+?		SIS	605	5	Normal; possible heterozygote; see GM3380 Lymphoid
GM3398	2	EEMEM 20U	0226	22	M	BL	+?		BRO	605	6	Normal; possible heterozygote; see GM3383 Lymphoid
*GM5823		EEMEM 20U	0424	18	M		--		PROBAND	868	1	AT5BI; increased gamma-ray induced sensitivity to cell killing; see GM5849 SV40 Transformed Fibroblast; passage 3 at IMR
							Bloom Syndrome - 21090					
*GM1492D	17	EEMEM 20U	0077	15	M	WH	--	A	PROBAND			B.S. Registry 844(AbRu); Ashkenazi; 46, XY; increased sister chromatid exchange and chromosome breakage in fibroblasts; increased post UV irradiation unscheduled DNA synthesis

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE								
Bloom Syndrome - 21090												
*GM1493D	12	EEMEM 20U	0077	4	M	WH	--	A	PROBAND			B.S. Registry #57(AmEl); Ashkenazi; 46, XY; increased sister chromatid exchange & chromosome breakage in fibroblasts; increased post UV irradiation unscheduled DNA synthesis
*GM2520A	11	EEMEM 20U	0178	10	F	WH	--	A	PROBAND			B.S. Registry #20(ViSh); 46,XX; increased frequency of sister chromatid exchange in fibroblasts
*GM2548A	4	EEMEM 20U	0070	6	M	BL	--	A	PROBAND			B.S. Registry #71(HaEn); American; 46, XY; increased frequency of sister chromatid exchange; increased post UV irradiation unscheduled DNA synthesis
*GM3509	13	EEMEM 20U	0044	3	M	WH	--	B	PROBAND			B.S. Registry #81(HaGrou), Line HG1219; French Canadian; increased breakage and sister chromatid exchange
*GM3510	8	EEMEM 20U	0044	20	M	WH	--	B	PROBAND			B.S. Registry #47(ArSmi), Line HG1251; Ashkenazi; increased chromosome breakage and sister chromatid exchange
*GM5289	9	EEMEM 20U	0335	5	F	OR	--	B	PROBAND			Japanese; B.S. Registry #86(NoKi); short stature, telangiectasias in face; IgA & IgM deficiency; lymphocytes & fibroblasts show increased sister chromatid exchange
*GM3402	3	EEMEM 20U	0110	30	M	WH	--		PROBAND	210	1	Ashkenazi; B.S. Registry #9(EmSh); congenital dwarf; photosensitive telangiectatic eruption of the face; see GM3403C Lymphoid
GM2085	19	EEMEM 20U	0044	21MO	F	WH	--	B	PROBAND	430	1	B.S. Registry #53(StAs); Ashkenazi; line HG916; increased frequency of sister chromatid exchange in fibroblasts; same patient as GM1620
*GM0811B	26	EEMEM 10I	0044	4	M	WH	--	A	PROBAND	500	1	B.S. Registry #3(HoCo); Ashkenazi; increased frequency of sister chromatid exchange in fibroblasts; same patient as GM2932
*GM2932	8	EEMEM 20U	0077	28	M	WH	--	A	PROBAND	500	1	B.S. Registry #3(HoCo); Ashkenazi; same patient as GM0811B; increased post UV irradiation unscheduled DNA synthesis

BIOCHEMICALLY MUTANT HUMAN FIBROBLASTS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Bloom Syndrome - 21090												
*GM34988	3	EEMEM 20U	0134	4	M	WH	--	B	PROBAND	720	1	B.S. Registry #87(AlFr); 46,XY; 14% of cells show random chromosome loss; excessive sister chromatid exchange in leukocytes & fibroblasts; see GM4408 Lymphoid
Fanconi Anemia (Pancytopenia) - 22765, 22766												
*GM0368A	10	EEMEM 20U	0133	8	M	BL	--	A	PROBAND			Multiple chromosome breaks; 46,XY; increased chromosome aberrations following treatment with mitomycin-C; clinically affected
*GM0369	11	EEMEM 10U	0133	6	M	WH	--		PROBAND			Multiple chromosome breaks and gaps; clinically affected
*GM0449A	12	EEMEM 10I	0044	6	F	WH	--		PROBAND			46,XX; classical symptoms; increased incidence of chromatid breaks & gaps following x-irradiation
*GM1746	5	EEMEM 20U	0077	15	M	WH	--		PROBAND			Arab; 46,XY in leukocytes with quadri & tri radials and breaks; in fibroblasts, 16% of cells show chromosome breaks; increased chromosome aberrations following treatment with mitomycin-C
*GM2061	8	EEMEM 20U	0133	20	F	WH	--		PROBAND			46,XX in leukocytes with chromosome breaks; several sibs also affected
*GM2361	8	EEMEM 15U	0188	14	M		--		PROBAND			Clinically affected
*GM0646	7	EEMEM 20U	0133	16	F	WH	--		PROBAND	365	1	Decreased colony-forming ability & cell survival following exposure to mitomycin C; clinically affected
GM2977	9	EEMEM 20U	0133	52	M	WH	+-(O)		FATHER	365	2	Clinically unaffected
GM2975	9	EEMEM 20U	0133	40	M	WH	+-(O)		PARENT	464	1	Father of affected child; proband not in Repository
GM2976	9	EEMEM 20U	0133	43	F	WH	+-(O)		PARENT	464	2	Mother of affected child; proband not in Repository
*GM2362	11	EEMEM 15U	0188	14	M		--		PROBAND	505	1	Similarly affected sib
GM2363	10	EEMEM 15U	0188	15	M		--		AFF BRO	505	2	Similarly affected sib
*GM1309	3	EEMEM 20U	0133	12	M	BL	--		PROBAND	969	1	Lymphocytes showed multiple chromosome breaks; clinically affected; high frequency of spontaneous chromosomal aberrations and extreme sensitivity to cell killing by mitomycin-C; see GM6914A SV40 Transformed Fibroblast

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Fanconi Anemia (Pancytopenia) - 22765, 22766												
*GM2053	4	EEMEM 20U	0077	12	F	WH	--	B	PROBAND	973	1	46,XX; multiple breaks, gaps, occasional dicentrics; Arab; clinically affected; cells are only slightly sensitive to cell killing by mitomycin-C
Incontinentia Pigmenti - 30830												
*GM0492	4	EEMEM 20U	0078	26	F		+-		PROBAND			Typical skin lesions appeared shortly after birth; 88% of fibroblasts are 46, XX; 10% show random chromosome loss; and 1 cell out of 50 showed t(12;14)
GM1236	9	EEMEM 20U	0109	3	F	WH	+-		PROBAND			46,XX; increased chromosome breakage in lymphocytes; normal (4-7%) in fibroblasts; clinically affected
GONADAL DYSGENESIS												
XX Male - 27885												
GM1889	3	EEMEM 20U	0125	24	M	WH		B	PROBAND			46,XX; phenotypically male; G6PD type B; Xg(a) antigen positive
*GM2670	7	R1640 15U	0030	19	M	WH		A	PROBAND	234	1	46,XX; phenotypically male; bilateral gynecomastia; negative for a segment of single-copy DNA (DXYS1) specific to the short arm of the Y chromosome in fibroblasts
*GM2624	12	R1640 20U	0030	45	M	WH			FATHER	234	2	Transient gynecomastia at age 20; in leukocytes, 2 cells out of 50 showed 46, XY,inv(7)(p15q36), remainder were normal
*GM2625	9	R1640 20U	0030	43	F	WH			MOTHER	234	3	46,XX in leukocytes; clinically normal
*GM2626	9	R1640 20U	0030	23	M	WH		B	AFF 2ND COU	234	4	46,XX; bilateral mild gynecomastia; average I.Q.; phenotypically male; no argininosuccinate synthetase-like Y chromosome DNA sequence or single-copy DNA (DXYS1) specific to the short arm of the Y chromosome in fibroblasts
*GM2671	7	R1640 15U	0030	47	F	WH			IN-LAW	234	5	46,XX in leukocytes and fibroblasts; clinically normal; mother of GM2626
*GM2672	11	R1640 20U	0030	48	M	WH			PAT COU	234	6	46,XY in leukocytes; clinically normal; father of GM2626
XY Female - 30610, 23342												
GM2600	5	EEMEM 20U	0109	31	F	WH		A	PROBAND			46,XY; H-Y antigen positive

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				GONADAL DYSGENESIS								
XY Female - 30610, 23342												
GM2601	6	EEMEM 20U	0109	28	F			A	PROBAND			46,XY; H-Y antigen positive; male pseudohermaphroditism; 10% of cells show random chromosome loss
GM1628	2	EEMEM 20U	0125	1	F	WH		A	PROBAND	122	1	46,XY; H-Y antigen positive; G6PD type B; Xg(a) antigen positive
GM1948	3	EEMEM 20U	0125	27	F	WH			MOTHER	122	2	Phenotypically normal; 46,XX,15p+
GM0048	7	EEMEM 20U	0095	1	F	WH		A	PROBAND	156	1	46,XY; right gonadal tissue, H-Y antigen ?; same patient as GM0083
GM0083	7	EEMEM 20U	0095	1	F	WH		A	PROBAND	156	1	46,XY; left gonadal tissue, H-Y antigen ?; same patient as GM0048
GM3368	4	EEMEM 20U	0172	17	F	BL		B	PROBAND	513	1	Pure gonadal dysgenesis; 46,XY; H-Y antigen ?; right gonadal tissue; same patient as GM3368A
GM3368A	6	EEMEM 20U	0172	17	F	BL		B	PROBAND	513	1	Pure gonadal dysgenesis; 46,XY; H-Y antigen ?; left gonadal streak; same patient as GM3368

Chromosomally Aberrant Human Fibroblasts

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAMG	FAMMG	REMARKS
TRANSLOCATION												
CHROMOSOME X												
*AG3499	2	EEMEM	20U 0195	11	F	WH	46,XX/46,X,t(X;5)(Xpter>Xq11::5q35>5qter;5pter>5q35::Xq11>Xqter)	B	PROBAND			Patient with basal cell nevus syndrome; treated with radio & chemotherapy; 85% of cells are 46,XX; mother has Charcot-Marie-Tooth disease and naevoid basal cell carcinoma
*GM0089	5	EEMEM	20U 0043		F	WH	46,X,t(X;19)(Xpter>Xq22::19q133>19qter;19pter>19q133::Xq22>Xqter)	B	A PROBAND			Normal X is late replicating
*GM0097	3	EEMEM	20U 0107	1	F	WH	46,X,t(X;1)(Xpter>Xq26::1q12>1qter;1pter>1q12::Xq26>Xqter)	B	A PROBAND			De novo translocation; Beckwith-Wiedemann syndrome; normal X is late replicating
GM0324	5	EEMEM	20U 0125	22	M	WH	47,XY,t(X;7)(Xpter>Xq24::7q32>7qter;7pter>7q32::Xq24>Xqter)	U	A PROBAND			Sardinian; homozygous for G6PD (med. type), Xg(a) antigen present
*GM0705	13	EEMEM	20U 0016	10MO	F	WH	46,X,t(X;9)(Xpter>Xq12::9p24>9pter;9qter>9p24::Xq12>Xqter)	B	A PROBAND			Patient with failure to thrive; culture is slow growing
*GM1695	5	EEMEM	20U 0176	26	F	WH	46,X,t(X;11)(Xqter>Xp2105::11q13>11qter;11pter>11q13::Xp2105>Xpter)	B	A PROBAND			Also has clinically documented Duchenne Muscular Dystrophy; in blood leukocytes and fibroblasts, normal X is late replicating
*GM1696A	9	EEMEM	20U 0176	30	F	WH	46,X,t(X;7)(Xpter>Xq21::7p22>7pter;7qter>7p22::Xq21>Xqter)	B	A PROBAND			Secondary amenorrhea; phenotypically normal; G6PD type B; normal X is late replicating; slow growing culture
GM1813	3	EEMEM	20U 0066	19	F	WH	46,X,t(X;15)(Xqter>Xp11::15q13>15qter;15pter>15q13::Xp11>Xpter),t(1;21)(q12q22)pat	B	A PROBAND			Menstrual dysfunction; t(X;15) present in 100% of cells at passage 6, but not seen in peripheral blood leukocytes
GM2103	6	EEMEM	20U 0109	19	F	WH	46,X,t(X;Y)(Xpter>Xq11::Yq11>Yqter)	U	A PROBAND			Secondary amenorrhea; abnormal X is late replicating
GM2469	2	EEMEM	20U 0070	17	F		46,X,t(X;Y)(Xpter>Xq22::Yq11>Yqter)	U	A PROBAND			Gonadal dysgenesis
*GM2621	8	EEMEM	10U 0224	32	F	WH	46,X,t(X;12)(Xpter>Xq22::12q24>12qter;12pter>12q24::Xq22>Xqter)	B	A PROBAND			Gonadal dysgenesis; positive for H-Y antigen; in blood leukocytes & fibroblasts the normal X chromosome was late replicating; elevated gonadotropins; bicornuate uterus; axillary & pubic hair; renal abnormalities

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME X												
GM2971	14	EEMEM	20U 0240	SMO	F	WH	46,X,t(X;13)(Xqter>Xp22::13q12>13qter; 13pter>13q12::Xp22>Xpter)	B	A	PROBAND		Sporadic Retinoblastoma; translocated X is the inactive X
*GM3322	4	EEMEM	20U 0070	35	F	WH	46,X,t(X;11)(Xpter>Xq22::11q13>11qter; 11pter>11q13::Xq22>Xqter)	B	A	PARENT		Phenotypically normal; normal menses; H-Y antigen negative; three offspring, one has the unbalanced translocation; normal X is late replicating
*GM3552	7	EEMEM	20U 0096	29	F	OR	46,X,t(X;11)(Xpter>Xq26::11q23> 11qter;11pter>11q23::Xq26>Xqter)	B	A	PROBAND		Vietnamese; gonadal dysgenesis; negative for H-Y antigen; in lymphocytes terminally pulsed with BrdU, normal X was late replicating; elevated gonadotropins; axillary and pubic hair
GM3884	7	EEMEM	10I 0354	14	F	WH	46,X,t(X;16)(Xpter>Xq26::16q24>16qter; 16pter>16q24::Xq26>Xqter)	B	A	PROBAND		Klippel-Feil syndrome; congenital heart murmur; bilaterally enlarged kidneys; mild hearing loss; inguinal hernias; normal X is late replicating; slow growing culture
GM4618	4	EEMEM	20U 0066	30	F	BL	46,X,t(X;1)(Xqter>Xp22.1::1q23>1qter; 1pter>1q23::Xp22.1>Xpter)	B	A	PARENT		A son has unbalanced translocation; normal X is late replicating
*GM4628	3	EEMEM	20U 0383		F		46,X,t(X;22)(Xpter>Xq12::22p11>22pter; 22qter>22p11::Xq12>Xqter)	B	A	PROBAND		Normal X is late replicating
*GM5835	6	EEMEM	20U 0426	10	F	OT	46,X,t(X;5)(Xqter>Xp21.2::5q35.3>5qter; 5pter>5q35.3::Xp21.2>Xpter)	B	A	PROBAND		Chinese-Hawaiian-Caucasian; clinically normal parents & 3 male sibs; short stature; elevated CPK; muscle atrophy & weakness; Duchenne muscular dystrophy; the normal X is late replicating; de novo translocation
GM7146	9	EEMEM	20I 0489	22	F	WH	46,X,t(X;10)(Xqter>Xp22.33::10q11.2> 10qter;10pter>10q11.2::Xp22.33>Xpter)	B	A	PROBAND		Mental retardation
GM7151	7	EEMEM	20I 0489	2WK	F	WH	46,X,t(X;17)(Xpter>Xq11.2::17q11.2> 17qter;17pter>17q11.2::Xq11.2>Xqter)	B	A	PROBAND		Multiple congenital defects including cleft palate, coloboma, tetralogy of Fallot, and abnormal genitalia and extremities

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME X												
*GM7503	12	EEMEM	20U 0506	10	F	WH	46,X,der(X),t(X;2)(Xpter>Xq28::2p21>2pter)mat	U	A PROBAND			Congenital heart disease; growth & mental retardation; alacrima; unusual triangular facies; strabismus; frontal bossing; simple, low set, posteriorly rotated ears; hypotonia
*GM7792	2	EEMEM	20U 0150	14	F	WH	46,X,t(X;20)(Xpter>cen>20qter;20pter>cen>Xqter)	B	A PROBAND			Mental retardation, seizures, & dysmorphic features; parents both have normal karyotypes; normal X is late replicating
*GM0439	6	EEMEM	20U 0069	41	F	WH	46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter)	B	B MOTHER	28	3	Unaffected mother of 2 children (GM0441 & GM0440) with galactosemia; 50% of normal transferase activity in erythrocytes; see GM1029 Lymphoid
*GM1533	7	EEMEM	20U 0069	19	F	WH	46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter)mat	B	B SIS	28	6	Clinically normal sib of galactosemia (transferase deficient) proband (GM0441); normal galactose-1-P-uridyl transferase and galactokinase activity in fibroblasts; normal X is late replicating
*GM1414	5	EEMEM	20U 0068	28	F	BL	46,X,-X,+der(9),t(X;9)(9pter>9q34::Xq13>Xqter)mat	U	A PROBAND	140	1	Turner's syndrome; in blood leukocytes and fibroblasts abnormal X is late replicating
*GM1429	8	EEMEM	20U 0068	55	F	BL	47,X,+10,t(X;9)(Xpter>Xq13::9q34>9qter;9pter>9q34::Xq13>Xqter)	U	A MOTHER	140	2	Mosaic; 16% of fibroblasts do not have additional 10; in blood leukocytes, normal X was late replicating; normal phenotype; fibroblasts have 164% of normal HK1 activity and 122% of normal GOT1 activity
*GM0074	10	EEMEM	10U 0111	23	M	WH	47,Y,t(X;14)(Xpter>Xq13::14q32>14qter;14pter>14q32::Xq13>Xqter),+der(14),t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A PROBAND	147	1	Klinefelter's syndrome; der(14) is late replicating in 88% of cells; trisomic for 14pter>14q32
*GM0073	5	EEMEM	20U 0111		F	WH	46,X,t(X;14)(Xpter>Xq13.1::14q32.1>14qter;14pter>14q32.1::Xq13.1>Xqter)	B	A MOTHER	147	2	Phenotypically normal; has had 6 spontaneous abortions; normal X is late replicating

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME X													
*GM1730	11	EEMEM	20U	0132	5	F	BL 46,XX,-21,+der(21),t(X;21)(21qter>21p11::Xq11>Xqter)mat	U	A	PROBAND	148	1	Mental retardation and multiple anomalies; one normal X and the translocated X are late replicating
*GM0194	15	EEMEM	20U	0030	19	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A	PROBAND	615	1	Clinically normal; Finnish; poor growing culture; in 20 metaphases, late replication studies showed 15 normal X chromosomes and 5 abnormal X chromosomes as late replicating; same pt. as GM2899
*GM2899	7	R1640	20U	0030	25	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A	PROBAND	615	1	Clinically normal; Finnish; same pt. as GM0194; in 158 metaphases, late replication studies showed 102 normal X chromosomes and 56 abnormal X chromosomes as late replicating
*GM2859	12	EEMEM	20U	0036	3	F	WH 46,X,del(11)(p11.2p1300),t(X;11)(Xpter>Xq11.1::11p1300>11pter;11qter>11p11.2::Xq11.1>Xqter)	U	A	PROBAND	617	1	See GM3316 Lymphoid; South American; in lymphoblast line, normal X is late replicating; mental retardation with bizarre dysmorphology syndrome: epicanthus, hypertelorism, & trigonocephaly
*GM5010	2	EEMEM	20U	0398	19	F	WH 46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2>2qter;2pter>2q14.2::8p11.1>8pter;8qter>8p11.1::Xq21.2>Xqter)	B	A	PROBAND	808	1	Primary amenorrhea; lack of 2ndary sexual devel.; no dysmorphic features; norm IQ; streak gonads & hypoplastic uterus; poss. del(X)(q21>q22.3); normal X is late rep. with 5% of cells showing label on trans. X seq.; see GM5011 Lymphoid
CHROMOSOME Y													
*GM0118	5	EEMEM	20U	0052	29	F	WH 46,XX,-15,+der(15),t(Y;15)(15qter>15p1::Yq11>Yqter)pat	U	A	PROBAND			Uterine tissue fibroblast culture; phenotypically normal
GM2103	6	EEMEM	20U	0109	19	F	WH 46,X,t(X;Y)(Xpter>Xq11::Yq11>Yqter)	U	A	PROBAND			Secondary amenorrhea; abnormal X is late replicating
GM2469	2	EEMEM	20U	0070	17	F	46,X,t(X;Y)(Xpter>Xq22::Yq11>Yqter)	U	A	PROBAND			Gonadal dysgenesis
*GM2598	3	EEMEM	20U	0109	18	F	WH 46,X,t(Y;17)(Ypter>Yq11::17p13>17pter;17qter>17p13::Yq11>Yqter)	B	A	PROBAND			Gonadoblastoma; streak gonad; amenorrhea; skin fibroblast culture

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 01													
*GM0097	3	EEMEM 20U	0107	1	F	WH	46,X,t(X;1)(Xpter>Xq26::1q12>1qter; 1pter>1q12::Xq26>Xqter)	B	A	PROBAND			De novo translocation; Beckwith-Wiedemann syndrome; normal X is late replicating
*GM0126	4	EEMEM 20U	0056	33	M	WH	46,XY,t(1;15)(1qter>1p36.1::15q11.2> 15qter;15pter>15q11.2::1p36.1>1pter)	B	A	PROBAND			Clinically normal
*GM0257	3	EEMEM 20U	0002	26	F		46,XX,t(1;2)(1pter>1q32::2p23>2pter; 2qter>2p23::1q32>1qter)	B	A	PROBAND			Phenotypically normal
*GM1356	2	EEMEM 20U	0066	26	F	WH	46,XX,t(1;7)(1qter>1p34::7p13>7pter; 7qter>7p13::1p34>1pter)	B	A	PROBAND			Phenotypically normal; history of repeated abortions; one phenotypically normal son with same translocation
*GM1421	12	EEMEM 20U	0109	53	M	BL	46,XY,t(1;6)(1pter>1q42::6q21>6qter; 6pter>6q21::1q42>1qter)	B	A	PROBAND			Phenotypically normal
GM1550	6	EEMEM 20U	0140	31	M	WH	46,XY,t(1;3;4)(1pter>1q32::3p21>3pter; 1qter>1q32::3p21>3q29::4p14>4pter; 4qter>4p14::3q29>3qter)	B	A	PROBAND			Family history of multiple abortions; wife is 46,XX
GM1564	9	EEMEM 20U	0151	28	F	WH	45,XX,-21,t(1;21)(1pter>1q44::21q11> 21qter)	U	A	PROBAND			Clinically normal
GM1813	3	EEMEM 20U	0066	19	F	WH	46,X,t(1;21)(1pter>1q12::21q22>21qter; 21pter>21q22::1q12>1qter)pat,t(X;15) (p11q13)	B	A	PROBAND			Menstrual dysfunction; t(X;15) present in 100% of cells at passage 6, but not seen in peripheral blood leukocytes
*GM1881	7	EEMEM 20U	0140	3	M	WH	46,XY,t(1;21)(1qter>1p31::21q22>21qter; 21pter>21q22::1p31>1pter)	B	A	PROBAND			Sanfilippo A; deficient heparan N-sulfatase; normal Alpha-N-acetylglucosaminidase activity
GM3137		EEMEM 20U	0237		F		46,XX/46,XX,t(1;12)(1qter>1p36::12q15> 12qter;12pter>12q15::1p36>1pter)	B	A	PROBAND			Adenosine deaminase deficiency with immunodeficiency; classical early onset, severe form; 52% of fibroblasts are 46, XX; passage 2 at IMR
GM3501	2	EEMEM 20U	0140	10	M	WH	46,XY,t(1;8)(1qter>1p34::8q22> 8qter;8pter>8q22::1p34>1pter)	B	A	PROBAND			Retardation-dysmorphic syndrome
GM3876	4	EEMEM 20U	0066	33	M	WH	46,XY,t(1;20)(1qter>1p13::20q133> 20qter;20pter>20q133::1p13>1pter)	B	A	PROBAND			Phenotypically normal

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							TRANSLOCATION						
CHROMOSOME 01													
GM4088	3	EEMEM 20U	0140	27	F	WH	46,XX,t(1;4)(1pter>1q32::4p16>4pter; 4qter>4p16::1q32>1qter)	B	A	PARENT			Two daughters with congenital anomalies died shortly after birth
GM4300	2	EEMEM 20U	0254	7	F	IN	46,XX,t(1;2)(1qter>1p32::2q11>2qter; 2pter>2q11::1p32>1pter)	B	A	PROBAND			Mental retardation & developmental delay; cannot rule out (p32;q13)
GM4500	8	EEMEM 20U	0078	15FW	F	WH	46,XX,t(1;3)(1qter>1p36::3q133>3qter; 3pter>3q133::1p36>1pter)	B	A	PROBAND			Spontaneous abortus
GM4618	4	EEMEM 20U	0066	30	F	BL	46,X,t(X;1)(Xqter>Xp22.1::1q23>1qter; 1pter>1q23::Xp22.1>Xpter)	B	A	PARENT			A son has unbalanced translocation; normal X is late replicating
*GM7457	4	EEMEM 20I	0504	24	F	WH	46,XX,t(1;21)(1pter>1q12::21q22.3> 21qter;21pter>21q22.3::1q12>1qter)	B	A	PROBAND			Healthy; has had two consecutive miscarriages at the 12th week of gestation
GM1229	3	EEMEM 20U	0007	3	F	BL	46,XX,-1,+der(1),t(1;2)(1qter>1p36:: 2q31>2qter)pat	U	A	PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM1230	2	EEMEM 20U	0007	44	M	BL	46,XY,t(1;2)(1qter>1p36::2q31>2qter; 2pter>2q31::1p36>1pter)	B	A	FATHER	129	2	Clinically normal
*GM3647	2	EEMEM 20U	0204	41	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25:: 1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1; similarly affected half-brother
*GM3648	2	EEMEM 20U	0204	32	M	WH	46,XY,-6,+der(6),t(1;6)(6qter>6p25:: 1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
*GM0201	10	EEMEM 20U	0030	13	F	WH	47,XX,+21,t(1;17)(1qter>1p32::17p13> 17pter;17qter>17p13::1p32>1pter)	U	A	PROBAND	337	1	Down's syndrome phenotype
GM3369	3	EEMEM 20U	0322	23	F	WH	46,XX,-1,+der(1),t(1;10)(1pter>1q44:: 10p11>10pter)mat	U	A	PROBAND	612	1	Developmental delay; similarly affected sister
GM3370	2	EEMEM 20U	0322	42	F	WH	46,XX,t(1;10)(1pter>1q44::10p11>10pter; 10qter>10p11::1q44>1qter)	B	A	MOTHER	612	2	Clinically normal; two daughters have the unbalanced translocation

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 01													
GM5347	2	EEMEM	20U 0074	2MO	M	WH	46,XY,-9,+der(9),t(1;9)(9qter>9p24::1q32>1qter)mat	U	A	PROBAND	849	1	Partial trisomy 1q; cup-shaped ear, small flattened nose, deep philtrum, downward slanting eyes, club feet, pyloric stenosis, VSD, and hyperconvex nails; mother is GM5348 Lymphoid
CHROMOSOME 02													
*GM0257	3	EEMEM	20U 0002	26	F		46,XX,t(1;2)(1pter>1q32::2p23>2pter;2qter>2p23::1q32>1qter)	B	A	PROBAND			Phenotypically normal
*GM0327	4	EEMEM	20U 0141	24	M	WH	46,XY,t(2;8)(2pter>2q13::8q24>8qter;8pter>8q24::2q13>2qter)	B	A	PARENT			Clinically normal; offspring had multiple congenital anomalies
GM1225	10	EEMEM	20U 0077	3MO	F	IN	46,XX,t(2;20)(2qter>2p21::20p13>20pter;20qter>20p13::2p21>2pter)	B	A	PROBAND			Asiatic Indian; de novo; has multiple congenital anomalies
GM1579	8	EEMEM	20U 0030	35	F	WH	46,XX,t(2;13)(2qter>2p13::13q32>13qter;13pter>13q32::2p13>2pter)	B	A	PROBAND			Multiple malformations & mental retardation
*GM40458	16	EEMEM	20U 0044	30MO	M	WH	46,XY,del(4)(q31.1q31.3),t(2;4)(2pter>2q14::4q31.3>4qter;4pter>4q31.1::2q14>2qter)	U	A	PROBAND			Mental retardation, facial dysmorphy, & unusual dermatoglyphics; hemizygous at MN blood group locus
GM4139	9	EEMEM	20U 0366	42	F	WH	46,XX,t(2;13)(2pter>2q31::13q32>13qter;13pter>13q32::2q31>2qter)	B	A	PARENT			Clinically normal; mother of a fetus with an unbalanced translocation
GM4300	2	EEMEM	20U 0254	7	F	IN	46,XX,t(1;2)(1qter>1p32::2q11>2qter;2pter>2q11::1p32>1pter)	B	A	PROBAND			Mental retardation & developmental delay; cannot rule out (p32;q13)
GM4613	4	EEMEM	20U 0107	1DA	M	WH	46,XY,t(2;11)(2qter>2p11::11p13>11pter;11qter>11p13::2p11>2pter)	B	A	PROBAND			Hypoplastic lungs; Potter facies; bilateral clubbed feet; bilateral undescended testes; kyphoscoliosis
*GM7503	12	EEMEM	20U 0506	10	F	WH	46,X,+der(X),t(X;2)(Xpter>Xq28::2p21>2pter)mat	U	A	PROBAND			Congenital heart disease; growth & mental retardation; alacrima; unusual triangular facies; strabismus; frontal bossing; simple, low set, posteriorly rotated ears; hypotonia

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 02												
GM1229	3	EEMEM	20U	0007	3	F	BL 46,XX,-1,+der(1),t(1;2)(1qter>1p36::2q31>2qter)pat	U	A PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM1230	2	EEMEM	20U	0007	44	M	BL 46,XY,t(1;2)(1qter>1p36::2q31>2qter;2pter>2q31::1p36>1pter)	B	A FATHER	129	2	Clinically normal
*GM0501	3	EEMEM	20U	0014	5	M	WH 46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A PROBAND	131	1	Growth and mental retardation
*GM1064	7	EEMEM	20U	0014	39	M	WH 46,XY,t(2;4)(2qter>2p25::4q21>4qter;4pter>4q21::2p25>2pter)	B	A FATHER	131	2	Phenotypically normal
*GM0845	7	EEMEM	20U	0018	18	F	WH 46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)mat	B	A PROBAND	132	1	Secondary amenorrhea subsequently resolved spontaneously; mother has no history of amenorrhea
*GM0846	8	EEMEM	20U	0018	54	F	WH 46,XX,t(2;8)(2pter>2q37::8q13>8qter;8pter>8q13::2q37>2qter)	B	A MOTHER	132	2	Multiple spontaneous abortions
*GM0692	5	EEMEM	20U	0001	8	M	WH 45,XY,-2,-21,+der(2),t(2;21)(2pter>2q37::21q2105>21qter)mat	U	A PROBAND	133	1	Mentally retarded
*GM0693	5	EEMEM	20U	0001	27	F	WH 46,XX,t(2;21)(2pter>2q37::21q2105>21qter;21pter>21q2105::2q37>2qter)	B	A MOTHER	133	2	Two Down's phenotype daughters; one mentally retarded son; and one normal child
GM1683	3	EEMEM	20U	0179	28	F	WH 46,XX,t(2;10)(2qter>2p23::10q26>10qter;10pter>10q26::2p23>2pter)	B	A PROBAND	134	1	Possible red cell acid phosphatase variant; slow growing culture
GM1848	3	EEMEM	20U	0179	35	M	WH 46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p23>2pter)mat	U	A MAT 1ST COU	134	2	Possible red cell acid phosphatase variant
GM1849	4	EEMEM	20U	0179	26	M	WH 46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p23>2pter)mat	U	A MAT 1ST COU	134	3	Possible red cell acid phosphatase variant; slow growing culture
GM2658	4	EEMEM	20U	0077	31	F	WH 46,XX,t(2;6)(2pter>2q11::6q15>6qter;6pter>6q15::2q11>2qter)	B	A PROBAND	242	1	Same patient as GM2699
*GM4371	3	EEMEM	20U	0354	4	M	OT 46,XY,-2,+der(2),t(2;5)(2qter>2p25::5q33>5qter)mat	U	A PROBAND	715	1	Black/Caucasian; multiple anomalies; partial trisomy 5q
GM4012	3	EEMEM	20U	0354	31	F	WH 46,XX,t(2;5)(2qter>2p25::5q33>5qter;5pter>5q33::2p25>2pter)	B	A MOTHER	715	2	Clinically normal

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 02													
*GM4409	2	EEMEM	20U 0204	6	M	WH	46,XY,-14,+der(14),t(2;14)(14pter> 14q32::2p23>2pter)mat	U	A	PROBAND	721	1	Mental retardation and congenital malformations
*GM4410	3	EEMEM	20U 0204	27	F	WH	46,XX,t(2;14)(2qter>2p23::14q32>14qter; 14pter>14q32::2p23>2pter)	B	A	MOTHER	721	2	Phenotypically normal; balanced carrier
*GM5010	2	EEMEM	20U 0398	19	F	WH	46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2> 2qter;2pter>2q14.2::8p11.1>8pter;8qter> 8p11.1::Xq21.2>Xqter)	B	A	PROBAND	808	1	Primary amenorrhea; lack of 2ndary sexual devel.; no dysmorphic features; norm IQ; streak gonads & hypoplastic uterus; poss. del(X)(q21>q22.3); normal X is late rep. with 5% of cells showing label on trans. X seq.; see GM5011 Lymphoid
CHROMOSOME 03													
GM1550	6	EEMEM	20U 0140	31	M	WH	46,XY,t(1;3;4)(1pter>1q32::3p21>3pter; 1qter>1q32::3p21>3q29::4p14>4pter; 4qter>4p14::3q29>3qter)	B	A	PROBAND			Family history of multiple abortions; wife is 46,XX
*GM2808	4	EEMEM	20U 0140	30	F	WH	46,XX,t(3;17)(3qter>3p21::17p13>17pter; 17qter>17p13::3p21>3pter)	B	A	PROBAND			Multiple spontaneous abortions
GM2860	10	EEMEM	20U 0036	42	M	WH	46,XY,t(3;16)(3pter>3q132::16q13>16qter; 16pter>16q13::3q132>3qter)	B	A	PROBAND			Family history of recurrent abortions
GM2963	3	EEMEM	20U 0247	3	M	BL	46,XY,t(3;17)(3qter>3p21::17q25>17qter; 17pter>17q25::3p21>3pter)	B	A	PROBAND			Unusual facies; hirsute; broad hands & feet; teeth overgrown with fibrous tissue
*GM3563	2	EEMEM	20U 0148	1MO	M	WH	46,XY,-9,+der(9),t(3;9)(9qter>9p24:: 3q12>3qter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM3577	2	EEMEM	20U 0107	8MO	M	WH	45,XY,-22,t(3;22)(3pter>3q29::22q11> 22qter)	U	A	PROBAND			DiGeorge syndrome; absent thymus and parathyroid glands; cleft lip and palate; congenital heart disease
GM3882	7	EEMEM	15U 0224	30	F	WH	46,XX,t(3;10)(3pter>3q25::10p15>10pter; 10qter>10p15::3q25>3qter)	B	A	PROBAND			Clinically normal with history of miscarriages
*GM4127	4	EEMEM	20U 0346	6MO	F	BL	46,XX,-12,+der(12),t(3;12)(12qter> 12p13::3p21>3pter)	U	A	PROBAND			Partial trisomy 3p; microcephaly; brachycephalic; cleft lip & palate
GM4500	8	EEMEM	20U 0078	15FW	F	WH	46,XX,t(1;3)(1qter>1p36::3q133>3qter; 3pter>3q133::1p36>1pter)	B	A	PROBAND			Spontaneous abortus

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAMS	FAMNO	REMARKS
TRANSLOCATION												
CHROMOSOME 03												
*GM5336	2	EEMEM	20U	0107	140A	M	WH 46,XY,-4,+der(4),t(3;4)(4qter>4p15::3p21.3>3pter)mat	U	A PROBAND			"Greek helmet face"; shallow orbits & protruding eyes; congenital heart disease; hypospadias; undescended testes
GM7216	5	EEMEM	20U	0224	25FW	F	WH 46,XX,-7,+der(7),t(3;7)(7pter>7q36::3p21.3>3pter)	U	A PROBAND			Cyclops; holoprosencephaly; proboscis; both parents have normal karyotypes
*GM0439	6	EEMEM	20U	0069	41	F	WH 46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter)	B	B MOTHER	28	3	Unaffected mother of 2 children (GM0441 & GM0440) with galactosemia; 50% of normal transferase activity in erythrocytes; see GM1029 Lymphoid
*GM1533	7	EEMEM	20U	0069	19	F	WH 46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter)mat	B	B SIS	28	6	Clinically normal sib of galactosemia (transferase deficient) proband (GM0441); normal galactose-1-P-uridyl transferase and galactokinase activity in fibroblasts; normal X is late replicating
AG3594		EEMEM	20U	0044	30	M	WH 46,XY,inv(3)(q23q29)+t(3;13)(p21;q32)+inv(9)(p11q34)	B	AFF BRO	546	2	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3593A; parents are first cousins
*GM0194	15	EEMEM	20U	0030	19	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A PROBAND	615	1	Clinically normal; Finnish; poor growing culture; in 20 metaphases, late replication studies showed 15 normal X chromosomes and 5 abnormal X chromosomes as late replicating; same pt. as GM2899
*GM2899	7	R1640	20U	0030	25	F	WH 46,X,t(X;3)(Xpter>Xq28::3q21>3qter;3pter>3q21::Xq28>Xqter)	B	A PROBAND	615	1	Clinically normal; Finnish; same pt. as GM0194; in 158 metaphases, late replication studies showed 102 normal X chromosomes and 56 abnormal X chromosomes as late replicating
CHROMOSOME 04												
*GM0098	5	EEMEM	20U	0119	10MO	M	46,XY,-(21),+der(21),t(4;21)(21qter>21p12::4p11>4pter)mat	U	A PROBAND			Multiple congenital anomalies
*GM0157	12	EEMEM	20U	0030	28	M	WH 49,XXXXY,t(4;11)(4pter>4q35::11q23>11qter;11pter>11q23::4q35>4qter)	U	A PROBAND			Severe mental retardation; sexual infantilism

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 04												
*GM0624	2	EEMEM	20U	0077	38	F	WH 46,XX,t(4;15)(4pter>4q11::15p11>15pter; 15qter>15p11::4q11>4qter)	B	A PROBAND			22% of cells show an additional t(6;11)(q13;q23) subline
*GM1091	3	EEMEM	20U	0058	27	F	WH 46,XX,t(4;10)(4pter>4q35::10q24>10qter; 10pter>10q24::4q35>4qter)	B	A PROBAND			History includes 1 miscarriage & 1 child with multiple congenital anomalies
GM1550	6	EEMEM	20U	0140	31	M	WH 46,XY,t(1;3;4)(1pter>1q32::3p21>3pter; 1qter>1q32::3p21>3q29::4p14>4pter; 4qter>4p14::3q29>3qter)	B	A PROBAND			Family history of multiple abortions; wife is 46,XX
GM3886	9	EEMEM	20U	0221	22	F	WH 46,XX,t(4;9)(4pter>4q33::9q12>9qter; 9pter>9q12::4q33>4qter)	B	A PROBAND			Clinically normal; has had five spontaneous first trimester abortions
*GM4045B	16	EEMEM	20U	0044	30MO	M	WH 46,XY,del(4)(q31.1q31.3),t(2;4)(2pter>2q14::4q31.3>4qter;4pter>4q31.1::2q14>2qter)	U	A PROBAND			Mental retardation, facial dysmorphy, & unusual dermatoglyphics; hemizygous at MN blood group locus
GM4088	3	EEMEM	20U	0140	27	F	WH 46,XX,t(1;4)(1pter>1q32::4p16>4pter; 4qter>4p16::1q32>1qter)	B	A PARENT			Two daughters with congenital anomalies died shortly after birth
GM4111	2	EEMEM	20U	0368	28	F	WH 46,XX,t(4;5)(4pter>4q31.1::5p15>5pter; 5qter>5p15::4q31.1>4qter),del(21)(qter>p11:)	U	A PARENT			Clinically normal; has 1 normal child; 1 child died & appeared to have Down's phenotype
GM4240	2	EEMEM	20U	0368	24	F	WH 46,XX,t(4;6)(4pter>4q25::6p25>6pter; 6qter>6p25::4q25>4qter)	B	A PROBAND			Patient has had 6 spontaneous abortions
GM4993	4	EEMEM	20U	0148	1MO	F	WH 46,XX,-10,+der(10),t(4;10)(10pter>10q26::4q31>4qter)mat	U	A PROBAND			Congenital heart disease, genito-urinary anomalies, malrotation of bowel, & dysmorphic features
*GM5336	2	EEMEM	20U	0107	140A	M	WH 46,XY,-4,+der(4),t(3;4)(4qter>4p15::3p21.3>3pter)mat	U	A PROBAND			"Greek helmet face"; shallow orbits & protruding eyes; congenital heart disease; hypospadias; undescended testes
GM5394	6	EEMEM	20U	0326	3MO	M	WH 46,XY,-4,+der(4),t(4;9)(4pter>4q35::9p21>9pter)mat	U	A PROBAND			Prominent forehead, low set ears, flat nasal bridge, high arched palate, micrognathia, short neck, bilateral simian creases, and hypoplastic toenails

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							TRANSLOCATION						
CHROMOSOME 04													
*GM5401	3	EEMEM	ZOU	0417	3MO	M	WH 45,XY,-4,-22,+der(4),t(4;22)(4pter>4q35::22q11>22qter)mat	U	A	PROBAND			Hispanic; partial DiGeorge syndrome; truncus arteriosis, type I; T-cell dysfunction; micrognathia and high arched palate; positive family history
*GM0501	3	EEMEM	ZOU	0014	5	M	WH 46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A	PROBAND	131	1	Growth and mental retardation
*GM1064	7	EEMEM	ZOU	0014	39	M	WH 46,XY,t(2;4)(2qter>2p25::4q21>4qter;4pter>4q21::2p25>2pter)	B	A	FATHER	131	2	Phenotypically normal
*GM1220	3	EEMEM	ZOU	0066	2	F	46,XX,-4,+der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A	PROBAND	136	1	Hypotonia, clubbed feet, and cardiac abnormality with cyanotic spells
*GM0773	5	EEMEM	ZOU	0066	26	F	46,XX,t(4;7)(4qter>4p16::7q34>7qter;7pter>7q34::4p16>4pter)	B	A	MOTHER	136	2	Clinically normal
*GM0380	2	EEMEM	ZOU	0078	35	F	WH 46,XX,t(4;11)(4pter>4q23::11q11>11qter;11pter>11q11::4q23>4qter)	B	A	PARENT	397	1	Clinically normal with a history of 2 normal children & 2 children who expired before age 1 month; see GM1561 Lymphoid
GM3811	4	EEMEM	ZOU	0354	56	F	WH 46,XX,t(4;14)(4qter>4p15::14p12>14pter;14qter>14p12::4p15>4pter)	B	A	MOTHER	552	1	Clinically normal; had 3 miscarriages & 1 stillborn
GM3810	4	EEMEM	101	0354	18	F	WH 46,XX,-14,+der(14),t(4;14)(14qter>14p12::4p15>4pter)mat	U	A	PROBAND	552	2	Partial trisomy 4p; mental and developmental retardation
*GM0972	3	EEMEM	ZOU	0067	13	F	BL 46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Mental retardation; see GM1063A Lymphoid
GM1101	7	EEMEM	ZOU	0007	32	M	WH 46,XY,t(4;12)(4qter>4p1608::12p11>12pter;12qter>12p11::4p1608>4pter)	B	A	FATHER	709	1	Clinically normal; balanced carrier
*GM1183	4	EEMEM	ZOU	0007	NB	M	WH 46,XY,-4,+der(4),t(4;12)(4qter>4p1608::12p11>12pter)pat	U	A	PROBAND	709	2	Small for dates; abnormal facies; coloboma; multiple anomalies
GM4921	4	EEMEM	ZOU	0140	31	F	WH 46,XX,t(4;8)(4qter>4p15::8p22>8pter;8qter>8p22::4p15>4pter)	B	A	PARENT	779	2	Phenotypically normal; two children with balanced and one with unbalanced translocation; proband not in Repository
GM5519	4	EEMEM	ZOU	0354	1	M	WH 46,XY,t(4;11)(4pter>4q25::11p13>11pter;11qter>11p13::4q25>4qter)mat	B	A	PROBAND	857	1	Bilateral vertical talus; epicanthal folds; mother is GM5654 Lymphoid; the sorted der(4) chromosome hybridized to gene specific probes for Insulin, ADJ-762, B-globin, PTH, & LDH-A

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 04													
GM7908	2	EEMEM	20U	0125		M	WH 46,XY,t(4;14)(4pter>4q25::14q32>14qter; 14pter>14q32::4q25>4qter)	B	A	PROBAND	1087	2	Daughter with same balanced translocation has had 3 spontaneous abortions
CHROMOSOME 05													
*AG3499	2	EEMEM	20U	0195	11	F	WH 46,XX/46,X,t(X;5)(Xpter>Xq11::5q35>5qter;5pter>5q35::Xq11>Xqter)	B		PROBAND			Patient with basal cell nevus syndrome; treated with radio & chemotherapy; 85% of cells are 46,XX; mother has Charcot-Marie-Tooth disease and naevoid basal cell carcinoma
*GM0071	15	EEMEM	20U	0015	19	F	WH 46,XX,-5,der(5),t(5;?)(p1;?)mat	U	A	PROBAND			Cri du Chat; deletion assumed to be derived rather than a simple deletion; ATCC CCL 90
*GM0344	4	EEMEM	20U	0066	7	M	WH 45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0589	2	EEMEM	20U	0012	18	M	WH 46,XY,t(5;14)(5qter>5p14::14q21>14qter; 14pter>14q21::5p14>5pter)	B	A	PROBAND			Clinically normal with emotional and mental disturbance
GM1678	3	EEMEM	20U	0140	33	F	46,XX,t(5;10)(5qter>5p15::10p13>10pter; 10qter>10p13::5p15>5pter)	B	A	PARENT			2 children with unbalanced translocation
*GM2426	5	EEMEM	20U	0134	12	M	WH 46,XY,t(5;6)(5qter>5p15::6p21>6pter; 6qter>6p21::5p15>5pter)	B	A	PROBAND			Clinically normal
GM4111	2	EEMEM	20U	0368	28	F	WH 46,XX,t(4;5)(4pter>4q31.1::5p15>5pter; 5qter>5p15::4q31.1>4qter),del(21)(qter>p11:)	U	A	PARENT			Clinically normal; has 1 normal child; 1 child died & appeared to have Down's phenotype
GM5409	3	EEMEM	20U	0016	28	M	WH 46,XY,t(5;17)(5pter>5q33::17p12>17pter; 17qter>17p12::5q33>5qter)	B	A	PARENT			Clinically normal; daughter with unbalanced translocation was multiply malformed
*GM5835	6	EEMEM	20U	0426	10	F	OT 46,X,t(X;5)(Xqter>Xp21.2::5q35.3>5qter; 5pter>5q35.3::Xp21.2>Xpter)	B	A	PROBAND			Chinese-Hawaiian-Caucasian; clinically normal parents & 3 male sibs; short stature; elevated CPK; muscle atrophy & weakness; Duchenne muscular dystrophy; the normal X is late replicating; de novo translocation

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 05													
GM6214	8	EEMEM	20U	0107	3	M	WH 46,XY,t(5;11)(5pter>5q22::11q23.1> 11qter;11pter>11q23.1::5q22>5qter)pat	B	A	PROBAND			Hurler syndrome; corneal clouding; developmental delay; coarse features; excess MPS in urine; deficient Alpha-L-Iduronidase and excess MPS accumulation in fibroblasts
GM3493	2	EEMEM	20U	0204	5MO	F	WH 46,XX,-5,+der(5),t(5;6)(5pter>5q35:: 6p22>6pter)pat	U	A	PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
GM3494A	5	EEMEM	20U	0204	27	M	WH 46,XY,t(5;6)(5pter>5q35::6p22>6pter; 6qter>6p22::5q35>5qter)	B	A	FATHER	135	2	Clinically normal
GM1535	6	EEMEM	20U	0002	16	F	BL 46,XX,-12,+der(12),t(5;12)(12pter> 12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM1536	6	EEMEM	20U	0002	38	F	BL 46,XX,t(5;12)(5pter>5q33::12q24>12qter; 12pter>12q24::5q33>5qter)	B	A	MOTHER	139	2	Balanced carrier
*GM2488		EEMEM	20U	0140	23	M	WH 46,XY,-15,+der(15),t(5;15)(15qter> 15p11::5p13>5qter)mat	U	A	PROBAND	225	1	Passage 6 at IMR
GM2487		EEMEM	20U	0140	44	F	WH 46,XX,t(5;15)(5qter>5p13::15p11>15pter; 15qter>15p11::5p13>5pter)	B	A	MOTHER	225	2	Passage 5 at IMR; translocation is present in all cells, different cells show different aberrations including X chromosome aberrations
*AG2651	3	EEMEM	20U	0195	46	M	WH 46,XY,t(5;8)(5qter>5p11::8q11>8qter; 8pter>8q11::5p11>5pter)	B		FATHER	308	5	Clinically normal; daughter has acute myelogenous leukemia (AG2655)
*AG3593A		EEMEM	20U	0044	28	M	WH 46,XY,t(5;8)(q11;q22),dup(6)(p21>p25), 9qh+	B		PROBAND	546	1	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3594; parents are first cousins
*GM3314	2	EEMEM	20U	0225	48	F	WH 46,XX/46,XX,t(5;15)(5pter>5q22::15q26> 15qter;15pter>15q26::5q22>5qter)	B		PROBAND	603	2	Gardner syndrome; 40% 46,XX; 14% of cells are polyploid at P5; see GM3315A Lymphoid
*GM3305	4	EEMEM	20U	0204	28	M	WH 46,XY,t(5;9)(5pter>5q33::9p24>9pter; 9qter>9p24::5q33>5qter)	B	A	PARENT	611	2	Clinically normal; proband not in Repository
*GM4371	3	EEMEM	20U	0354	4	M	OT 46,XY,-2,+der(2),t(2;5)(2qter>2p25:: 5q33>5qter)mat	U	A	PROBAND	715	1	Black/Caucasian; multiple anomalies; partial trisomy 5q

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 05													
GM4012	3	EEMEM	20U	0354	31	F	WH 46,XX,t(2;5)(2qter>2p25::5q33>5qter; 5pter>5q33::2p25>2pter)	B	A	MOTHER	715	2	Clinically normal
CHROMOSOME 06													
*GM0144	5	EEMEM	20U	0012	34	F	WH 46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A	PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture; Down's syndrome; 3 male sibs have balanced translocation
*GM0610	7	EEMEM	20U	0075	11WK	F	WH 46,XX,t(6;18)(6pter>6q21::18p11>18pter; 18qter>18p11::6q21>6qter)	B	A	PROBAND			Phenotypically normal
*GM1137	6	R1640	10U	0017	1MO	M	BL 48,XXY,+21,t(6;10)(6qter>6p25::10p11> 10pter;10qter>10p11::6p25>6pter)	U	A	PROBAND			Down's syndrome features
*GM1139	4	EEMEM	20U	0095	21	F	WH 46,XX,t(6;13)(6qter>6p21::13q34>13qter; 13pter>13q34::6p21>6pter),t(15;17)(q15; p13)	B	A	PARENT			Culture is mosaic: at passage 6, 64% carry only the t(15;17) & 36% carry both; mother of a severely defective child with the unbalanced translocation
*GM1421	12	EEMEM	20U	0109	53	M	BL 46,XY,t(1;6)(1pter>1q42::6q21>6qter; 6pter>6q21::1q42>1qter)	B	A	PROBAND			Phenotypically normal
*GM1605	3	EEMEM	20U	0166	34	M	WH 46,XY,t(6;11)(6qter>6p23::11q23>11qter; 11pter>11q23::6p23>6pter)	B	A	PROBAND			Phenotypically normal with a family history of infertility and spontaneous abortions
*GM2068	5	EEMEM	10U	0088	4	F	WH 46,XX,t(6;7)(6pter>6q27::7q22>7qter; 7pter>7q22::6q27>6qter)	B	A	PROBAND			
*GM2426	5	EEMEM	20U	0134	12	M	WH 46,XY,t(5;6)(5qter>5p15::6p21>6pter; 6qter>6p21::5p15>5pter)	B	A	PROBAND			Clinically normal
GM2594	4	EEMEM	20U	0073	30	M	WH 46,XY,t(6;9)(6pter>6q13::9p22>9pter; 9qter>9p22::6q13>6qter)	B	A	PROBAND			Clinically normal; wife (46,XX) has history of several spontaneous abortions
GM3404	2	EEMEM	20U	0066	28	F	46,XX,t(6;12)(6pter>6q21::12p13>12pter; 12qter>12p13::6q21>6qter)	B	A	PROBAND			
GM4240	2	EEMEM	20U	0368	24	F	WH 46,XX,t(4;6)(4pter>4q25::6p25>6pter; 6qter>6p25::4q25>4qter)	B	A	PROBAND			Patient has had 6 spontaneous abortions
GM7069A	4	EEMEM	20U	0390	22FW	F	WH 46,XX,t(6;10)(6pter>6q25.3::10q26.1> 10qter;10pter>10q26.1::6q25.3>6qter)	B	A	PROBAND			A twin fetus (dizygotic) has the unbalanced translocation

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CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 06													
GM3493	2	EEMEM	20U	0204	5MO	F	WH 46,XX,-5,+der(5),t(5;6)(5pter>5q35::6p22>6pter)pat	U	A	PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
GM3494A	5	EEMEM	20U	0204	27	M	WH 46,XY,t(5;6)(5pter>5q35::6p22>6pter;6qter>6p22::5q35>5qter)	B	A	FATHER	135	2	Clinically normal
GM2658	4	EEMEM	20U	0077	31	F	WH 46,XX,t(2;6)(2pter>2q11::6q15>6qter;6pter>6q15::2q11>2qter)	B	A	PROBAND	242	1	Same patient as GM2699
*GM3647	2	EEMEM	20U	0204	41	M	WH 46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1; similarly affected half-brother
*GM3648	2	EEMEM	20U	0204	32	M	WH 46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
*GM4188	3	EEMEM	20U	0367	77	F	WH 46,XX/46,XX,t(6;19)(6pter>6q13::19p13 3>19pter;19qter>19p13 3::6q13>6qter)	B		SPOUSE	682	2	Clinically normal; 78% of cells are 46,XX; see GM4189 Lymphoid; husband (GM4219) has Huntington's Disease
GM5183A	4	EEMEM	20U	0039	49	M	WH 46,XY,t(6;7)(6pter>6q21::7q21.2>7qter;7pter>7q21.2::6q21>6qter)	B	A	PROBAND	827	1	Wife has had three 1st trimester spontaneous abortions; see GM5184 Lymphoid
GM5655	2	EEMEM	20U	0204	1MO	M	WH 46,XY,-21,+der(21),t(6;21)(21qter>21p11.2::6q22.2>6qter)mat	U	A	PROBAND	858	1	Multiple congenital anomalies and dysmorphic features
GM5656	2	EEMEM	20U	0204	26	F	WH 46,XX,t(6;21)(6pter>6q22.2::21p11.2>21pter;21qter>21p11.2::6q22.2>6qter)	B	A	MOTHER	858	2	Clinically normal; balanced carrier
CHROMOSOME 07													
*GM0044	5	EEMEM	20U	0097	6	M	WH 46,XY,del(7)(p21.2p22.1),t(7;10)(7qter>7p21.2::10q11.21>10qter;10pter>10q11.21::7p22.1>7pter)	U	A	PROBAND			Congenital malformations, severe retardation
GM0324	5	EEMEM	20U	0125	22	M	WH 47,XY,t(X;7)(Xpter>Xq24::7q32>7qter;7pter>7q32::Xq24>Xqter)	U	A	PROBAND			Sardinian; homozygous for G6PD (med. type), Xg(a) antigen present
*GM0657	3	EEMEM	20U	0107	4WK	M	WH 46,XY,del(7)(q34q36),t(7;18)(7pter>7q34::18q12.2>18qter;18pter>18q12.2::7q36>7qter)mat	U	A	PROBAND			Cleft palate; microcephaly; developmental delay; similarly affected cousin; clinically normal mother has balanced t(7;18) in lymphocytes

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 07													
*GM1356	2	EEMEM	20U	0066	26	F	WH 46,XX,t(1;7)(1qter>1p34::7p13>7pter; 7qter>7p13::1p34>1pter)	B	A	PROBAND			Phenotypically normal; history of repeated abortions; one phenotypically normal son with same translocation
*GM1696A	9	EEMEM	20U	0176	30	F	WH 46,X,t(X;7)(Xpter>Xq21::7p22>7pter; 7qter>7p22::Xq21>Xqter)	B	A	PROBAND			Secondary amenorrhea; phenotypically normal; G6PD type B; normal X is late replicating; slow growing culture
*GM2068	5	EEMEM	10U	0088	4	F	WH 46,XX,t(6;7)(6pter>6q27::7q22>7qter; 7pter>7q22::6q27>6qter)	B	A	PROBAND			
GM3118	2	EEMEM	20U	0320		F	46,XX,del(11)(p13p11)/46,XX,del(11) (p13p11),t(7;22)(p15;p12)	U	A	PROBAND			Bilateral Wilms tumor and bilateral aniridia; 40% of cells do not show the translocation
GM4862	2	EEMEM	20U	0315	24	F	WH 46,XX,t(7;21)(7pter>7q36::21q21> 21qter;21pter>21q21::7q36>7qter)	B	A	PROBAND			Clinically normal
GM6097A	3	EEMEM	20U	0417	15MO	F	WH 46,XX,-17,+der(17),t(7;17)(17qter> 17p13.2::7p22.3>7pter)pat	U	A	PROBAND			Lissencephaly syndrome (Miller-Dieker syndrome); typical facies; ventricular-septal defect
GM7216	5	EEMEM	20U	0224	25FW	F	WH 46,XX,-7,+der(7),t(3;7)(7pter>7q36:: 3p21.3>3pter)	U	A	PROBAND			Cyclops; holoprosencephaly; proboscis; both parents have normal karyotypes
*GM1220	3	EEMEM	20U	0066	2	F	46,XX,-4,+der(4),t(4;7)(4qter>4p16:: 7q34>7qter)mat	U	A	PROBAND	136	1	Hypotonia, clubbed feet, and cardiac abnormality with cyanotic spells
*GM0773	5	EEMEM	20U	0066	26	F	46,XX,t(4;7)(4qter>4p16::7q34>7qter; 7pter>7q34::4p16>4pter)	B	A	MOTHER	136	2	Clinically normal
GM1840	7	EEMEM	20U	0031	10	F	WH 45,XX,-13,t(7;13)(7qter>7p22::13q12.1> 13qter)	U	A	PROBAND	183	1	Mental retardation; hemihypertrophy - left side; developmental delay; fibroblast culture from right arm skin biopsy
GM5183A	4	EEMEM	20U	0039	49	M	WH 46,XY,t(6;7)(6pter>6q21::7q21.2>7qter; 7pter>7q21.2::6q21>6qter)	B	A	PROBAND	827	1	Wife has had three 1st trimester spontaneous abortions; see GM5184 Lymphoid
GM5382	8	EEMEM	20U	0134	27	F	WH 46,XX,t(7;18)(7pter>7q36::18q12.2> 18qter;18pter>18q12.2::7q36>7qter)	B	A	PARENT	851	2	Clinically normal; proband not in Repository; unbalanced child showed mental retardation, developmental delay, seizures, dysmorphic facial features, syndactyly, and optic atrophy

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 08													
*GM0213	5	EEMEM	20U 0082	35	M	WH	46,XY,t(8;12)(8qter>8p23::12p11>12pter; 12qter>12p11::8p23>8pter)	B	A	PARENT			Clinically normal; son has clinical features resembling Down's syndrome
*GM0327	4	EEMEM	20U 0141	24	M	WH	46,XY,t(2;8)(2pter>2q13::8q24>8qter; 8pter>8q24::2q13>2qter)	B	A	PARENT			Clinically normal; offspring had multiple congenital anomalies
GM1512	6	EEMEM	20U 0030	26	M	WH	46,XY,t(8;10)(8qter>8p21::10p15>10pter; 10qter>10p15::8p21>8pter)	B	A	PROBAND			Small testes
GM3501	2	EEMEM	20U 0140	10	M	WH	46,XY,t(1;8)(1qter>1p34::8q22> 8qter;8pter>8q22::1p34>1pter)	B	A	PROBAND			Retardation-dysmorphic syndrome
*GM3689	6	EEMEM	20U 0247	24	F	WH	46,XX,-10,+der(10),t(8;10)(10qter> 10p15::8p21.1>8pter)mat	U	A	PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
*GM0345	7	EEMEM	20U 0018	18	F	WH	46,XX,t(2;8)(2pter>2q37::8q13>8qter; 8pter>8q13::2q37>2qter)mat	B	A	PROBAND	132	1	Secondary amenorrhea subsequently resolved spontaneously; mother has no history of amenorrhea
*GM0846	8	EEMEM	20U 0018	54	F	WH	46,XX,t(2;8)(2pter>2q37::8q13>8qter; 8pter>8q13::2q37>2qter)	B	A	MOTHER	132	2	Multiple spontaneous abortions
*AG2651	3	EEMEM	20U 0195	46	M	WH	46,XY,t(5;8)(5qter>5p11::8q11>8qter; 8pter>8q11::5p11>5pter)	B		FATHER	308	5	Clinically normal; daughter has acute myelogenous leukemia (AG2655)
*GM1920	2	EEMEM	20U 0061	23	M	WH	47,XY,+21,inv(9)(pter>p11::q13>p11:: q13>qter)mat,t(8;14)(8pter>8q11::14q11> 14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	Down's syndrome phenotype; see GM1921A Lymphoid
*AG3593A		EEMEM	20U 0044	28	M	WH	46,XY,t(5;8)(q11;q22),dup(6)(p21>p25), 9qh+	B		PROBAND	546	1	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3594; parents are first cousins
*GM3255	3	EEMEM	20U 0204	3MO	M	WH	46,XY,-15,+der(15),t(8;15)(15pter> 15q26::8p21>8pter)mat	U	A	PROBAND	609	1	Partial trisomy 8(p); growth retardation; dysmorphic features; skeletal abnormalities; congenital heart defect
*GM3296	3	EEMEM	20U 0204	25	F	WH	46,XX,t(8;15)(8qter>8p21::15q26>15qter; 15pter>15q26::8p21>8pter)	B	A	MOTHER	609	2	Clinically normal; balanced carrier

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAMB	FAMM	REMARKS
							TRANSLOCATION					
CHROMOSOME 08												
GM4921	4	EEMEM	20U 0140	31	F	WH	46,XX,t(4;8)(4qter>4p15::8p22>8pter; 8qter>8p22::4p15>4pter)	B	A PARENT	779	2	Phenotypically normal; two children with balanced and one with unbalanced translocation; proband not in Repository
*GM5010	2	EEMEM	20U 0398	19	F	WH	46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2> 2qter;2pter>2q14.2::8p11.1>8pter;8qter> 8p11.1::Xq21.2>Xqter)	B	A PROBAND	808	1	Primary amenorrhea; lack of 2ndary sexual devel.; no dysmorphic features; norm IQ; streak gonads & hypoplastic uterus; poss. del(X)(q21>q22.3); normal X is late rep. with 5% of cells showing label on trans. X seg.; see GM5011 Lymphoid
CHROMOSOME 09												
*GM0344	4	EEMEM	20U 0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14 ::15q11>15qter),t(9;11)(9qter>9p24:: 11q23>11qter;11pter>11q23::9p24>9pter)	U	A PROBAND			Cri du Chat
*GM0705	13	EEMEM	20U 0016	10MO	F	WH	46,X,t(X;9)(Xpter>Xq12::9p24>9pter; 9qter>9p24::Xq12>Xqter)	B	A PROBAND			Patient with failure to thrive; culture is slow growing
*GM1664	6	EEMEM	20U 0113	40	M	WH	46,XY,t(9;13)(9pter>9q13::13q12>13qter; 13pter>13q12::9q13>9qter)	B	A PROBAND			Phenotypically normal
GM1892	5	EEMEM	20U 0132	37	F	WH	46,XX,t(9;18)(9qter>9p24::18q12> 18qter;18pter>18q12::9p24>9pter)	B	A PROBAND			
*GM2492	6	EEMEM	20U 0221	38	M	WH	46,XY,t(9;13)(9qter>9p22 or p23::13q14 or q21>13qter;13pter>13q14 or q21::9p22 or p23>9pter)	B	A PROBAND			Phenotypically normal; family history of recurrent abortions
GM2594	4	EEMEM	20U 0073	30	M	WH	46,XY,t(6;9)(6pter>6q13::9p22>9pter; 9qter>9p22::6q13>6qter)	B	A PROBAND			Clinically normal; wife (46,XX) has history of several spontaneous abortions
*GM2836	4	EEMEM	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17) (q12;p11),ins(3;1)(q21;q25q44)	B	A PROBAND			Clinically normal; 61% of cells do not have the insertion (3;1)
GM3268	2	EEMEM	20U 0122	10MO	M	BL	46,XY,-21,+t(9;21)(9pter>cen>21qter)	U	A PROBAND			Clinodactyly; short neck; bulbous nose; low set ears
*GM3563	2	EEMEM	20U 0148	1MO	M	WH	46,XY,-9,+der(9),t(3;9)(9qter>9p24:: 3q12>3qter)mat	U	A PROBAND			Multiple congenital anomalies
GM3886	9	EEMEM	20U 0221	22	F	WH	46,XX,t(4;9)(4pter>4q33::9q12>9qter; 9pter>9q12::4q33>4qter)	B	A PROBAND			Clinically normal; has had five spontaneous first trimester abortions

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 09													
GM5394	6	EEMEM	20U	0326	3MO	M	WH 46,XY,-4,+der(4),t(4;9)(4pter>4q35::9p21>9pter)mat	U	A	PROBAND			Prominent forehead, low set ears, flat nasal bridge, high arched palate, micrognathia, short neck, bilateral simian creases, and hypoplastic toenails
*GM1414	5	EEMEM	20U	0068	28	F	BL 46,X,-X,+der(9),t(X;9)(9pter>9q34::Xq13>Xqter)mat	U	A	PROBAND	140	1	Turner's syndrome; in blood leukocytes and fibroblasts abnormal X is late replicating
*GM1429	8	EEMEM	20U	0068	55	F	BL 47,X,+10,t(X;9)(Xpter>Xq13::9q34>9qter;9pter>9q34::Xq13>Xqter)	U	A	MOTHER	140	2	Mosaic: 16% of fibroblasts do not have additional 10; in blood leukocytes, normal X was late replicating; normal phenotype; fibroblasts have 164% of normal HK1 activity and 122% of normal GOT1 activity
*GM1750	2	EEMEM	20U	0070	30MO	M	WH 47,XY,+der(14),t(9;14)(14pter>14q21::9p24>9pter)mat	U	A	PROBAND	141	1	Three first cousins have the same partial trisomy 14; growth retardation, facial dysplasia, & developmental delay
*GM1751	2	EEMEM	20U	0070	26	F	WH 46,XX,t(9;14)(9qter>9p24::14q21>14qter;14pter>14q21::9p24>9pter)	B	A	MOTHER	141	2	Four sisters have same translocation
GM3462	2	EEMEM	20U	0204	3MO	F	WH 47,XX,+der(14),t(9;14)(14pter>14q13::9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations
GM3463	4	EEMEM	20U	0204	23	F	WH 46,XX,t(9;14)(9qter>9p22::14q13>14qter;14pter>14q13::9p22>9pter)	B	A	MOTHER	227	2	Clinically normal; balanced carrier
GM1734	3	EEMEM	20U	0140	40	M	46,XY,-15,+der(9),t(9;15)(9pter>9q12::15q11>15qter)mat	U	A	PROBAND	257	1	
GM2819	3	EEMEM	20U	0107	7MO	F	BL 46,XX,-12,+der(12),t(9;12)(12pter>12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
GM2821	2	EEMEM	20U	0107		F	BL 46,XX,t(9;12)(9qter>9p13::12q24>12qter;12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2822 Lymphoid; balanced carrier
*GM1387	4	EEMEM	20U	0061	14	F	WH 46,XX,-13,+der(9),t(9;13)(9pter>9q22::13q12>13qter)mat or 46,XX,-13,+der(9),t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid; profoundly retarded; clinical features of trisomy 9p syndrome

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMNO	REMARKS
TRANSLOCATION													
CHROMOSOME 09													
GM3119	4	EEMEM	20U	0172	28	F	WH 46,XX,t(9;17)(9qter>9p22::17p11>17pter; 17qter>17p11::9p22>9pter)	B	A	MOTHER	449	2	Fetus is GM3100 Amniotic; history of several spontaneous abortions
*GM3305	4	EEMEM	20U	0204	28	M	WH 46,XY,t(5;9)(5pter>5q33::9p24>9pter; 9qter>9p24::5q33>5qter)	B	A	PARENT	611	2	Clinically normal; proband not in Repository
*GM0988	10	EEMEM	10U	0082	3WK	M	WH 46,XY,t(9;17)(9qter>9p13::17q25>17qter; 17pter>17q25::9p13>9pter)	B	A	PROBAND	702	1	Multiple congenital anomalies; 13% of cells are polyploid
GM5347	2	EEMEM	20U	0074	2MO	M	WH 46,XY,-9,+der(9),t(1;9)(9qter>9p24::1q32>1qter)mat	U	A	PROBAND	849	1	Partial trisomy 1q; cup-shaped ear, small flattened nose, deep philtrum, downward slanting eyes, club feet, pyloric stenosis, VSD, and hyperconvex nails; mother is GM5348 Lymphoid
*GM6033	1	EEMEM	20U	0107	5	F	BL 46,XX,-12,+der(12),t(9;12)(12pter>12q24.31::9p13>9pter)mat	U	A	PROBAND	998	1	Congenital heart disease, loose joints, and unusual facies
GM7548	6	EEMEM	20U	0107	12FW	F	BL 46,XX,t(9;12)(9qter>9p13::12q24.31> 12qter;12pter>12q24.31::9p13>9pter)mat	B	A	AFF COU	998	3	Fetal resorbtion; maternal cousin of proband; mothers are sisters; culture established from fetal villi
*GM5319	4	EEMEM	20U	0107	24FW	F	WH 47,XX,+der(9),t(9;13)(9pter>9q31.2::13q32.1>13qter)mat	U	A	PROBAND	1042	1	Mother with balanced translocation is GM7456 Lymphoid
CHROMOSOME 10													
*GM0044	5	EEMEM	20U	0097	6	M	WH 46,XY,del(7)(p21.2p22.1),t(7;10)(7qter> 7p21.2::10q11.21>10qter;10pter>10q11.21 ::7p22.1>7pter)	U	A	PROBAND			Congenital malformations, severe retardation
*GM1091	3	EEMEM	20U	0058	27	F	WH 46,XX,t(4;10)(4pter>4q35::10q24>10qter; 10pter>10q24::4q35>4qter)	B	A	PROBAND			History includes 1 miscarriage & 1 child with multiple congenital anomalies
*GM1137	6	R1640	10U	0017	1MO	M	BL 48,XXY,+21,t(6;10)(6qter>6p25::10p11> 10pter;10qter>10p11::6p25>6pter)	U	A	PROBAND			Down's syndrome features
*GM1396	3	EEMEM	20U	0095	3	M	WH 46,XY,-10,+der(10),t(10;16)(10pter> 10q26::16q22>16qter)pat	U	A	PROBAND			Mental retardation; no speech and questionable hearing; clinically normal father's leukocytes show 46,XY,t(10;16)(q26;q22)

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 10													
GM1512	6	EEMEM	20U 0030	26	M	WH	46,XY,t(8;10)(8qter>8p21i::10p15>10pter; 10qter>10p15::8p21i>8pter)	B	A	PROBAND			Small testes
GM1678	3	EEMEM	20U 0140	33	F		46,XX,t(5;10)(5qter>5p15::10p13>10pter; 10qter>10p13::5p15>5pter)	B	A	PARENT			2 children with unbalanced translocation
GM3241A	3	EEMEM	20U 0016	10MO	F	WH	45,XX,-10,-13,+der(10),t(10;13)(10pter> 10q26::13q12>13qter)	U	A	PROBAND			Multiple congenital anomalies; both parents have normal karyotypes
*GM3689	6	EEMEM	20U 0247	24	F	WH	46,XX,-10,+der(10),t(8;10)(10qter> 10p15::8p21.1>8pter)mat	U	A	PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
GM3882	7	EEMEM	15U 0224	30	F	WH	46,XX,t(3;10)(3pter>3q25::10p15>10pter; 10qter>10p15::3q25>3qter)	B	A	PROBAND			Clinically normal with history of miscarriages
GM4993	4	EEMEM	20U 0148	1MO	F	WH	46,XX,-10;+der(10),t(4;10)(10pter> 10q26::4q31>4qter)mat	U	A	PROBAND			Congenital heart disease, genito-urinary anomalies, malrotation of bowel, & dysmorphic features
GM7069A	4	EEMEM	20U 0390	22FW	F	WH	46,XX,t(6;10)(6pter>6q25.3::10q26.1> 10qter;10pter>10q26.1::6q25.3>6qter)	B	A	PROBAND			A twin fetus (dizygotic) has the unbalanced translocation
GM7146	9	EEMEM	20I 0489	22	F	WH	46,X,t(X;10)(Xqter>Xp22.33::10q11.2> 10qter;10pter>10q11.2::Xp22.33>Xpter)	B	A	PROBAND			Mental retardation
GM1683	3	EEMEM	20U 0179	28	F	WH	46,XX,t(2;10)(2qter>2p23::10q26> 10qter;10pter>10q26::2p23>2pter)	B	A	PROBAND	134	1	Possible red cell acid phosphatase variant; slow growing culture
GM1848	3	EEMEM	20U 0179	35	M	WH	46,XY,-10,+der(10),t(2;10)(10pter> 10q26::2p23>2pter)mat	U	A	MAT 1ST COU	134	2	Possible red cell acid phosphatase variant
GM1849	4	EEMEM	20U 0179	26	M	WH	46,XY,-10,+der(10),t(2;10)(10pter> 10q26::2p23>2pter)mat	U	A	MAT 1ST COU	134	3	Possible red cell acid phosphatase variant; slow growing culture
*GM0217	6	EEMEM	20U 0143	9DA	M	WH	46,XY,-17,+der(17),t(10;17)(17qter> 17p13::10q24>10qter)pat	U	A	PROBAND	142	1	Culture grows slowly; multiple anomalies
*GM0216	4	EEMEM	20U 0143	29	M	WH	46,XY,t(10;17)(10pter>10q24::17p13> 17pter;17qter>17p13::10q24>10qter)	B	A	FATHER	142	2	Clinically normal; balanced carrier

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							TRANSLOCATION						
CHROMOSOME 10													
*GM0959	6	EEMEM 20U	0143	11	M	WH	46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)mat	U	A	MAT 3RD COU	142	3	Multiple congenital anomalies; growth & mental retardation; increased soluble glutamic oxaloacetic transaminase activity
*GM1413	8	EEMEM 20U	0132	18MO	F	BL	46,XX,-10,+der(10),t(10;21)(10pter>10q26::21q2105>21qter)mat	U	A	PROBAND	143	1	Down's syndrome
*GM1580	6	EEMEM 20U	0132	48	F	BL	46,XX,t(10;21)(10pter>10q26::21q21>21qter;21pter>21q21::10q26>10qter)	B	A	MAT GRMR	143	2	Normal phenotype
*GM1399	5	EEMEM 20U	0132	20	M	BL	47,XY,+der(21),t(10;21)(21pter>21q2105::10q26>10qter)mat	U	A	MAT 1ST COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM0983	3	EEMEM 20U	0095	14MO	M	WH	46,XY,-21,+der(21),t(10;21)(21pter>21q22::10q24>10qter)mat	U	A	PROBAND	144	1	Normal soluble glutamic oxaloacetic transaminase activity
*GM0984B	8	EEMEM 20U	0095	34	F	WH	46,XX,t(10;21)(10pter>10q24::21q22>21qter;21pter>21q22::10q24>10qter)	B	A	MOTHER	144	2	Balanced carrier
GM3369	3	EEMEM 20U	0322	23	F	WH	46,XX,-1,+der(1),t(1;10)(1pter>1q44::10p11>10pter)mat	U	A	PROBAND	612	1	Developmental delay; similarly affected sister
GM3370	2	EEMEM 20U	0322	42	F	WH	46,XX,t(1;10)(1pter>1q44::10p11>10pter;10qter>10p11::1q44>1qter)	B	A	MOTHER	612	2	Clinically normal; two daughters have the unbalanced translocation
CHROMOSOME 11													
*GM0084A	8	EEMEM 20U	0095	1MO	M	WH	47,XY,+der(22),t(11;22)(22pter>22q11::11q23>11qter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM0157	12	EEMEM 20U	0030	28	M	WH	49,XXXXY,t(4;11)(4pter>4q35::11q23>11qter;11pter>11q23::4q35>4qter)	U	A	PROBAND			Severe mental retardation; sexual infantilism
*GM0344	4	EEMEM 20U	0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0980	2	EEMEM 20U	0012	33	F	WH	45,XX,-11,-22,+der(11),t(1?;22)(11pter>11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)
*GM1605	3	EEMEM 20U	0166	34	M	WH	46,XY,t(6;11)(6qter>6p23::11q23>11qter;11pter>11q23::6p23>6pter)	B	A	PROBAND			Phenotypically normal with a family history of infertility and spontaneous abortions

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 11												
*GM1695	5	EEMEM	20U 0176	26	F	WH	46,X,t(X;11)(Xqter>Xp2105::11q13>11qter; 11pter>11q13::Xp2105>Xpter)	B	A PROBAND			Also has clinically documented Duchenne Muscular Dystrophy; in blood leukocytes and fibroblasts, normal X is late replicating
*GM3005	8	EEMEM	20U 0268	28	F	WH	46,XX,t(11;16)(11pter>11q13::16p11> 16pter;16qter>16p11::11q13>11qter)	B	A PROBAND			Phenotypically normal; mother and daughter have same balanced translocation
*GM3322	4	EEMEM	20U 0070	35	F	WH	46,X,t(X;11)(Xpter>Xq22::11q13>11qter; 11pter>11q13::Xq22>Xqter)	B	A PARENT			Phenotypically normal; normal menses; H-Y antigen negative; three offspring, one has the unbalanced translocation; normal X is late replicating
*GM3552	7	EEMEM	20U 0096	29	F	OR	46,X,t(X;11)(Xpter>Xq26::11q23> 11qter;11pter>11q23::Xq26>Xqter)	B	A PROBAND			Vietnamese; gonadal dysgenesis; negative for H-Y antigen; in lymphocytes terminally pulsed with BrdU, normal X was late replicating; elevated gonadotropins; axillary and pubic hair
GM3847	8	EEMEM	15U 0353	38	F	WH	46,XX,t(11;22)(11pter>11q25::22q13> 22qter;22pter>22q13::11q25>11qter)mat	B	A PROBAND			Clinically normal; mother has same translocation; neither shows any myeloproliferative disorder
GM4613	4	EEMEM	20U 0107	1DA	M	WH	46,XY,t(2;11)(2qter>2p11::11p13>11pter; 11qter>11p13::2p11>2pter)	B	A PROBAND			Hypoplastic lungs; Potter facies; bilateral clubbed feet; bilateral undescended testes; kyphoscoliosis
GM6214	8	EEMEM	20U 0107	3	M	WH	46,XY,t(5;11)(5pter>5q22::11q23.1> 11qter;11pter>11q23.1::5q22>5qter)pat	B	A PROBAND			Hurler syndrome; corneal clouding; developmental delay; coarse features; excess MPS in urine; deficient Alpha-L-Iduronidase and excess MPS accumulation in fibroblasts
GM7332	2	EEMEM	20U 0012	1DA	M	WH	46,XY,t(11;22)(11pter>11q23::22q11.2> 22qter;22pter>22q11.2::11q23>11qter)mat	B	A PROBAND			Placenta biopsy; mother has same balanced translocation
*GM0380	2	EEMEM	20U 0078	35	F	WH	46,XX,t(4;11)(4pter>4q23::11q11>11qter; 11pter>11q11::4q23>4qter)	B	A PARENT	397	1	Clinically normal with a history of 2 normal children & 2 children who expired before age 1 month; see GM1561 Lymphoid

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
TRANSLOCATION													
CHROMOSOME 11													
*GM3371	2	EEMEM	20U 0322	5WK	F	BL	47,XX, +der(22),t(11;22)(22pter>22q11::11q23>11qter)mat, inv(9)(p13q21)pat	U	A	PROBAND	613	1	Partial trisomy 22; developmental delay
GM3372	2	EEMEM	20U 0322	28	F	BL	46,XX,t(11;22)(11pter>11q23::22q11>22qter;22pter>22q11::11q23>11qter)	B	A	MOTHER	613	2	Clinically normal
*GM2859	12	EEMEM	20U 0036	3	F	WH	46,X,del(11)(p11.2p1300),t(X;11)(Xpter>Xq11.1::11p1300>11pter;11qter>11p11.2::Xq11.1>Xqter)	U	A	PROBAND	617	1	See GM3316 Lymphoid; South American; in lymphoblast line, normal X is late replicating; mental retardation with bizarre dysmorphology syndrome: epicanthus, hypertelorism, & trigonocephaly
*GM4370A	3	EEMEM	20U 0007	4	F	WH	47,XX,+der(22),t(11;22)(22pter>22q11::11q23>11qter)mat	U	A	PROBAND	718	1	Microcephaly; earpits; micrognathia; mentally retarded
GM4403	4	EEMEM	20U 0007	30	F	WH	46,XX,t(11;22)(11pter>11q23::22q11>22qter;22pter>22q11::11q23>11qter)	B	A	MOTHER	718	2	Clinically normal
GM5519	4	EEMEM	20U 0354	1	M	WH	46,XY,t(4;11)(4pter>4q25::11p13>11pter;11qter>11p13::4q25>4qter)mat	B	A	PROBAND	857	1	Bilateral vertical talus; epicanthal folds; mother is GM5654 Lymphoid; the sorted der(4) chromosome hybridized to gene specific probes for insulin, ADJ-762, B-globin, PTH, & LDH-A
CHROMOSOME 12													
*GM0213	5	EEMEM	20U 0082	35	M	WH	46,XY,t(8;12)(8qter>8p23::12p11>12pter;12qter>12p11::8p23>8pter)	B	A	PARENT			Clinically normal; son has clinical features resembling Down's syndrome.
GM1665	3	EEMEM	20U 0140	28	F	WH	46,XX,t(12;21)(12qter>12p11::21p11>21pter;21qter>21p11::12p11>12pter)	B	A	PROBAND			Clinically normal
*GM2621	8	EEMEM	10U 0224	32	F	WH	46,X,t(X;12)(Xpter>Xq22::12q24>12qter;12pter>12q24::Xq22>Xqter)	B	A	PROBAND			Gonadal dysgenesis; positive for H-Y antigen; in blood leukocytes & fibroblasts the normal X chromosome was late replicating; elevated gonadotropins; bicornuate uterus; axillary & pubic hair; renal abnormalities
GM3137		EEMEM	20U 0237		F		46,XX/46,XX,t(1;12)(1qter>1p36::12q15>12qter;12pter>12q15::1p36>1pter)	B	A	PROBAND			Adenosine deaminase deficiency with immunodeficiency; classical early onset, severe form; 52% of fibroblasts are 46, XX; passage 2 at IMR

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#S	REMARKS
							TRANSLOCATION						
CHROMOSOME 12													
GM3404	2	EEMEM 20U	0066	28	F		46,XX,t(6;12)(6pter>6q21::12p13>12pter; 12qter>12p13::6q21>6qter)	B	A	PROBAND			
*GM4127	4	EEMEM 20U	0346	6MO	F	BL	46,XX,-12,+der(12),t(3;12)(12qter> 12p13::3p21>3pter)	U	A	PROBAND			Partial trisomy 3p; microcephaly; brachycephalic; cleft lip & palate
GM1535	6	EEMEM 20U	0002	16	F	BL	46,XX,-12,+der(12),t(5;12)(12pter> 12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM1536	6	EEMEM 20U	0002	38	F	BL	46,XX,t(5;12)(5pter>5q33::12q24>12qter; 12pter>12q24::5q33>5qter)	B	A	MOTHER	139	2	Balanced carrier
*AG3607	2	EEMEM 20U	0195	44	F	WH	46,XX/46,XX,t(12;13)(12qter>12p11:: 13q34>13qter;13pter>13q34::12p11>12pter)	B		MOTHER	308	4	Patient has had a cystadenoma of the ovary & invasive carcinoma of the cervix which was treated with irradiation; 56% of fibroblasts are 46,XX; son (AG2655) has acute myelogenous leukemia
GM2819	3	EEMEM 20U	0107	7MO	F	BL	46,XX,-12,+der(12),t(9;12)(12pter> 12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
GM2821	2	EEMEM 20U	0107		F	BL	46,XX,t(9;12)(9qter>9p13::12q24> 12qter;12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2822 Lymphoid; balanced carrier
GM3950	4	EEMEM 20U	0225	12	F	WH	46,XX/46,XX,t(12;13)(12pter>12q11:: 13q34>13qter;13pter>13q34::12q11>12qter)	B		DAU	603	8	At risk for Gardner syndrome; mother (GM3314) and five sibs are affected; see GM3951A Lymphoid; 10% of cells are 46,XX
GM1101	7	EEMEM 20U	0007	32	M	WH	46,XY,t(4;12)(4qter>4p1608::12p11> 12pter;12qter>12p11::4p1608>4pter)	B	A	FATHER	709	1	Clinically normal; balanced carrier
*GM1183	4	EEMEM 20U	0007	NB	M	WH	46,XY,-4,+der(4),t(4;12)(4qter> 4p1608::12p11>12pter)pat	U	A	PROBAND	709	2	Small for dates; abnormal facies; coloboma; multiple anomalies
*GM6033	1	EEMEM 20U	0107	5	F	BL	46,XX,-12,+der(12),t(9;12)(12pter> 12q24.31::9p13>9pter)mat	U	A	PROBAND	998	1	Congenital heart disease, loose joints, and unusual facies
GM7548	6	EEMEM 20U	0107	12FW	F	BL	46,XX,t(9;12)(9qter>9p13::12q24.31> 12qter;12pter>12q24.31::9p13>9pter)mat	B	A	AFF COU	998	3	Fetal resorbtion; maternal cousin of proband; mothers are sisters; culture established from fetal villi
CHROMOSOME 13													
*GM0085	6	EEMEM 20U	0143	2WK	M	BL	46,XY,-13,+t(13;13)(13qter>cen>13qter)	U	A	PROBAND			Clinical Trisomy 13

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#2	REMARKS
							TRANSLOCATION						
CHROMOSOME 13													
*GM0392	9	EEMEM	ZOU	0002	1	F	WH 45,XX,t(13;22)(13qter>cen>22qter)	U	A	PROBAND			Myelomonocytic leukemia
*GM0627	3	EEMEM	ZOU	0058	30	F	BL 46,XX,t(13;22)(13pter>13q22::22q13>22qter;22pter>22q13::13q22>13qter)	B	A	PARENT			Clinically normal; son has multiple anomalies and unbalanced translocation
*GM1139	4	EEMEM	ZOU	0095	21	F	WH 46,XX,t(6;13)(6qter>6p21::13q34>13qter;13pter>13q34::6p21>6pter),t(15;17)(q15;p13)	B	A	PARENT			Culture is mosaic: at passage 6, 64% carry only the t(15;17) & 36% carry both; mother of a severely defective child with the unbalanced translocation
*GM1224	7	EEMEM	ZOU	0077	3MO	F	WH 46,XX,-13,+der(13),t(13;18)(13pter>13q34::18q11>18qter)pat	U	A	PROBAND			Multiple congenital anomalies
GM1296	4	EEMEM	ZOU	0012	47	F	WH 45,XX,t(13;15)(13qter>cen>15qter)	U	A	PROBAND			Clinically normal; in addition to t(13;15) some cells show 21p+ and 10q- which could be a t(10;21)(q24;p12)
GM1579	8	EEMEM	ZOU	0030	35	F	WH 46,XX,t(2;13)(2qter>2p13::13q32>13qter;13pter>13q32::2p13>2pter)	B	A	PROBAND			Multiple malformations & mental retardation
*GM1664	6	EEMEM	ZOU	0113	40	M	WH 46,XY,t(9;13)(9pter>9q13::13q12>13qter;13pter>13q12::9q13>9qter)	B	A	PROBAND			Phenotypically normal
*GM2018	12	EEMEM	ZOU	0077	2	F	WH 45,XX,tan(13;13)(pter>q34::q12>qter)	U	A	PROBAND			Delayed psychomotor development; multiple anomalies; hyperkinesis
*GM2492	6	EEMEM	ZOU	0221	38	M	WH 46,XY,t(9;13)(9qter>9p22 or p23::13q14 or q21>13qter;13pter>13q14 or q21::9p22 or p23>9pter)	B	A	PROBAND			Phenotypically normal; family history of recurrent abortions
GM2813	7	R1640	ZOU	0148	1WK	F	WH 46,XX,-15,+t(13;15)(13qter>cen>15qter)mat	U	A	PROBAND			Multiple anomalies; polydactyly; trisomy 13q
GM2971	14	EEMEM	ZOU	0240	5MO	F	WH 46,X,t(X;13)(Xqter>Xp22::13q12>13qter;13pter>13q12::Xp22>Xpter)	B	A	PROBAND			Sporadic Retinoblastoma; translocated X is the inactive X
GM3044	3	EEMEM	ZOU	0148	2WK	A	WH 45,XY,t(13;14)(13qter>cen>14qter)	U	A	PROBAND			Smith-Lemli-Opitz syndrome; clinically affected

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							TRANSLOCATION						
CHROMOSOME 13													
GM5241A	3	EEMEM	20U	0016	10MO	F	WH 45,XX,-10,-13,+der(10),t(10;13)(10pter> 10q26::13q12>13qter)	U	A	PROBAND			Multiple congenital anomalies; both parents have normal karyotypes
GM3786	6	EEMEM	20U	0353	19	F	WH 45,XX,dic(13;14)(13qter>13p11::14p11> 14qter)	U	A	PROBAND			Greek; clinically normal
GM4139	9	EEMEM	20U	0366	42	F	WH 46,XX,t(2;13)(2pter>2q31::13q32>13qter; 13pter>13q32::2q31>2qter)	B	A	PARENT			Clinically normal; mother of a fetus with an unbalanced translocation
GM4890	2	EEMEM	20U	0368	32	F	WH 45,XX,dic(13;14)(13qter>13p11::14p11> 14qter)	U	A	PROBAND			Clinically normal; has had 5 miscarriages
*GM5832	4	EEMEM	20U	0425	20FW	M	WH 46,XY,-18,+der(18),t(13;18)(18pter> 18q23::13q14.1>13qter)	U	A	PROBAND			Partial trisomy 13; multiple external & internal malformations including brachycephaly, bilateral epicanthus, malformed ears, hexadactyly, vitium cordis, & cystic kidneys
GM7147	7	EEMEM	20I	0489	31	F	46,XX,t(13;16)(13pter>13q12.1::16p13.11> 16pter;16qter>16p13.11::13q12.1>13qter)	B	A	PROBAND			Clinically normal
GM7150	5	EEMEM	20I	0489	18FW	F	47,XX,+der(13),t(13;17)(13pter>13q22:: 17q25>17qter)mat	U	A	PROBAND			
*GM1663	13	EEMEM	20U	0113	14	M	WH 46,XY,-17,+der(17),t(13;17)(17qter> 17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13; severe mental and motor retardation
*GM1555	5	EEMEM	20U	0113	13	M	WH 47,XY,+der(13),t(13;17)(13pter>13q14:: 17p13>17pter)mat	U	A	BRO	145	2	Mental retardation; minor dysmorphia
GM1840	7	EEMEM	20U	0031	10	F	WH 45,XX,-13,t(7;13)(7qter>7p22::13q12.1> 13qter)	U	A	PROBAND	183	1	Mental retardation; hemihypertrophy - left side; developmental delay; fibroblast culture from right arm skin biopsy
GM3089	6	EEMEM	20U	0299	3WK	F	WH 46,XX,-18,+der(18),t(13;18)(18pter> 18q23::13q31>13qter)pat	U	A	PROBAND	301	1	Anomalies consistent with both trisomy 13 & 18
*AG3607	2	EEMEM	20U	0195	44	F	WH 46,XX/46,XX,t(12;13)(12qter>12p11:: 13q34>13qter;13pter>13q34::12p11>12pter)	B		MOTHER	308	4	Patient has had a cystadenoma of the ovary & invasive carcinoma of the cervix which was treated with irradiation; 56% of fibroblasts are 46,XX; son (AG2655) has acute myelogenous leukemia

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 13													
*GM1387	4	EEMEM	20U 0061	14	F	WH	46,XX,-13,+der(9),t(9;13)(9pter>9q22::13q12>13qter)mat or 46,XX,-13,+der(9),t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid; profoundly retarded; clinical features of trisomy 9p syndrome
AG3594		EEMEM	20U 0044	30	M	WH	46,XY,inv(3)(q23q29),t(3;13)(p21;q32),inv(9)(p11q34)	B		AFF BRO	546	2	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3593A; parents are first cousins
GM3950	4	EEMEM	20U 0225	12	F	WH	46,XX/46,XX,t(12;13)(12pter>12q11::13q34>13qter;13pter>13q34::12q11>12qter)	B		DAU	603	8	At risk for Gardner syndrome; mother (GM3314) and five sibs are affected; see GM3951A Lymphoid; 10% of cells are 46,XX
*GM0972	3	EEMEM	20U 0067	13	F	BL	46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Mental retardation; see GM1063A Lymphoid
GM6526	2	EEMEM	20U 0107	4WK	M	BL	45,XY,-22,t(13;22)(13pter>13q34::22q11.2>22qter)	U	A	PROBAND	959	1	Skin biopsy; unusual facies: small jaw, flat philtrum, downturned upper lip, cupped ears, & sloping forehead with low hairline; unusually long fingers with distal phalanges angled upwards
*GM5319	4	EEMEM	20U 0107	24FW	F	WH	47,XX,+der(9),t(9;13)(9pter>9q31.2::13q32.1>13qter)mat	U	A	PROBAND	1042	1	Mother with balanced translocation is GM7456 Lymphoid
CHROMOSOME 14													
*GM0005	5	EEMEM	20U 0111	30	F	WH	45,XX,t(14;22)(14qter>cen>22qter)	U	A	PROBAND			Clinically normal
*GM0479	12	EEMEM	20U 0048	35	F	WH	45,XX,t(14;15)(14qter>cen>15qter)pat	U	A	PROBAND			Four spontaneous abortions; father has t(14;15)
*GM0589	2	EEMEM	20U 0012	18	M	WH	46,XY,t(5;14)(5qter>5p14::14q21>14qter;14pter>14q21::5p14>5pter)	B	A	PROBAND			Clinically normal with emotional and mental disturbance
GM2044	2	EEMEM	20U 0164	7	M		45,XY,-16,t(14;16)(16;18)(14qter>14p12::16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			Slight macrocephaly; generalized hypotonia; mongoloid slant to eyes
GM3044	3	EEMEM	20U 0148	2WK	A	WH	45,XY,t(13;14)(13qter>cen>14qter)	U	A	PROBAND			Smith-Lemli-Opitz syndrome; clinically affected

REP #	PAS #	CULTURE MEDIA	SMTK CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 14													
GM3417	5	EEMEM	20U	0324	15	F	WH 45,XX,t(14;21)(14qter>cen>21qter)/ 46,XX,-14,+t(14;21)(14qter>cen>21qter)	U	A	PROBAND			Down's syndrome mosaic; 32% of cells are balanced 45,XX,t(14;21)
GM3786	6	EEMEM	20U	0353	19	F	WH 45,XX,dic(13;14)(13qter>13p11::14p11> 14qter)	U	A	PROBAND			Greek; clinically normal
GM4610		EEMEM	20U	0080		F	46,XX,dic(14;21)(14qter>14p13::21p13> 21qter),+8/47,XX,+8	U	A	PROBAND			Additional t(14;21) present in only 25% of cells at passage 6; passage 3 at IMR
GM4890	2	EEMEM	20U	0368	32	F	WH 45,XX,dic(13;14)(13qter>13p11::14p11> 14qter)	U	A	PROBAND			Clinically normal; has had 5 miscarriages
*GM1750	2	EEMEM	20U	0070	30MO	M	WH 47,XY,+der(14),t(9;14)(14pter>14q21:: 9p24>9pter)mat	U	A	PROBAND	141	1	Three first cousins have the same partial trisomy 14; growth retardation, facial dysplasia, & developmental delay
*GM1751	2	EEMEM	20U	0070	26	F	WH 46,XX,t(9;14)(9qter>9p24::14q21> 14qter;14pter>14q21::9p24>9pter)	B	A	MOTHER	141	2	Four sisters have same translocation
*GM0981	3	EEMEM	20U	0148	4	F	BL 46,XX,-14,+der(14),t(14;20)(14qter> 14p11.2::20p11.1>20pter)mat,inv(9)(pter> p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	Retardation and multiple anomalies
*GM0982	3	EEMEM	20U	0148		F	BL 46,XX,t(14;20)(14qter>14p11.2::20p11.1> 20pter;20qter>20p11.1::14p11.2>14pter), inv(9)(p12q12)	B	A	MOTHER	146	2	Phenotypically normal; balanced carrier
GM5133	3	EEMEM	20U	0148	FW	M	BL 46,XY,-14,+der(14),t(14;20)(14qter> 14p11.2::20p11.1>20pter)mat	U	A	AFF BRO	146	3	Abortus
*GM0074	10	EEMEM	10U	0111	23	M	WH 47,Y,t(X;14)(Xpter>Xq13::14q32>14qter; 14pter>14q32::Xq13>Xqter),+der(14), t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A	PROBAND	147	1	Klinefelter's syndrome; der(14) is late replicating in 88% of cells; trisomic for 14pter>14q32
*GM0073	5	EEMEM	20U	0111		F	WH 46,X,t(X;14)(Xpter>Xq13.1::14q32.1> 14qter;14pter>14q32.1::Xq13.1>Xqter)	B	A	MOTHER	147	2	Phenotypically normal; has had 6 spontaneous abortions; normal X is late replicating
GM3462	2	EEMEM	20U	0204	3MO	F	WH 47,XX,+der(14),t(9;14)(14pter>14q13:: 9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 14													
GM3463	4	EEMEM 20U	0204	23	F	WH	46,XX,t(9;14)(9qter>9p22::14q13>14qter; 14pter>14q13::9p22>9pter)	B	A	MOTHER	227	2	Clinically normal; balanced carrier
*GM1920	2	EEMEM 20U	0061	23	M	WH	47,XY,+21,inv(9)(pter>p11::q13>p11:: q13>qter)mat,t(8;14)(8pter>8q11::14q11> 14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	Down's syndrome phenotype; see GM1921A Lymphoid
GM3811	4	EEMEM 20U	0354	56	F	WH	46,XX,t(4;14)(4qter>4p15::14p12> 14pter;14qter>14p12::4p15>4pter)	B	A	MOTHER	552	1	Clinically normal; had 3 miscarriages & 1 stillborn
GM3810	4	EEMEM 10I	0354	18	F	WH	46,XX,-14,+der(14),t(4;14)(14qter> 14p12::4p15>4pter)mat	U	A	PROBAND	552	2	Partial trisomy 4p; mental and developmental retardation
*GM4409	2	EEMEM 20U	0204	6	M	WH	46,XY,-14,+der(14),t(2;14)(14pter> 14q32::2p23>2pter)mat	U	A	PROBAND	721	1	Mental retardation and congenital malformations
*GM4410	3	EEMEM 20U	0204	27	F	WH	46,XX,t(2;14)(2qter>2p23::14q32>14qter; 14pter>14q32::2p23>2pter)	B	A	MOTHER	721	2	Phenotypically normal; balanced carrier
GM7908	2	EEMEM 20U	0125		M	WH	46,XY,t(4;14)(4pter>4q25::14q32>14qter; 14pter>14q32::4q25>4qter)	B	A	PROBAND	1087	2	Daughter with same balanced translocation has had 3 spontaneous abortions
CHROMOSOME 15													
*GM0017	4	EEMEM 20U	0012	30MO	M	BL	45,XY,-15,-18,+der(18),t(15;18)(18pter> 18q23::15q13>15qter)	U	A	PROBAND			Multiple congenital anomalies & mental retardation
*GM0118	5	EEMEM 20U	0052	29	F	WH	46,XX,-15,+der(15),t(Y;15)(15qter>15p1:: Yq11>Yqter)pat	U	A	PROBAND			Uterine tissue fibroblast culture; phenotypically normal
*GM0126	4	EEMEM 20U	0056	33	M	WH	46,XY,t(1;15)(1qter>1p36.1::15q11.2> 15qter;15pter>15q11.2::1p36.1>1pter)	B	A	PROBAND			Clinically normal
*GM0344	4	EEMEM 20U	0066	7	M	WH	45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14 ::15q11>15qter),t(9;11)(9qter>9p24:: 11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
*GM0479	12	EEMEM 20U	0048	35	F	WH	45,XX,t(14;15)(14qter>cen>15qter)pat	U	A	PROBAND			Four spontaneous abortions; father has t(14;15)
*GM0624	2	EEMEM 20U	0077	38	F	WH	46,XX,t(4;15)(4pter>4q11::15p11>15pter; 15qter>15p11::4q11>4qter)	B	A	PROBAND			22% of cells show an additional t(6;11)(q13;q23) subline

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 15												
*GM1139	4	EEMEM	20U 0095	21	F	WH	46,XX,t(15;17)(15pter>15q15::17p13>17pter;17qter>17p13::15q15>15qter),t(6;13)(p21;q34)	B	A PARENT			Culture is mosaic: at passage 6, 64% carry only the t(15;17) & 36% carry both; mother of a severely defective child with the unbalanced translocation
GM1296	4	EEMEM	20U 0012	47	F	WH	45,XX,t(13;15)(13qter>cen>15qter)	U	A PROBAND			Clinically normal; in addition to t(13;15) some cells show 21p+ and 10q- which could be a t(10;21)(q24;p12)
GM1613	3	EEMEM	20U 0066	19	F	WH	46,X,t(X;15)(Xqter>Xp11::15q13>15qter;15pter>15q13::Xp11>Xpter),t(1;21)(q12q22)pat	B	A PROBAND			Menstrual dysfunction; t(X;15) present in 100% of cells at passage 6, but not seen in peripheral blood leukocytes
GM2729	2	EEMEM	20U 0116	15	M	WH	47,XY,+t(15;15)(15pter>15q13::15p11>15pter)	U	A PROBAND			
GM2813	7	R1640	20U 0148	1WK	F	WH	46,XX,-15,+t(13;15)(13qter>cen>15qter)mat	U	A PROBAND			Multiple anomalies; polydactyly; trisomy 13q
*GM4125	5	EEMEM	20U 0340	1DA	M	WH	46,XY,t(15;18)(15pter>15q15::18p13>18pter;18qter>18p13::15q15>15qter)	B	A PROBAND			Phenotypically normal; died 4 hrs after birth
*GM2488		EEMEM	20U 0140	23	M	WH	46,XY,-15,+der(15),t(5;15)(15qter>15p11::5p13>5pter)mat	U	A PROBAND	225	1	Passage 6 at IMR
GM2487		EEMEM	20U 0140	44	F	WH	46,XX,t(5;15)(5qter>5p13::15p11>15pter;15qter>15p11::5p13>5pter)	B	A MOTHER	225	2	Passage 5 at IMR; translocation is present in all cells, different cells show different aberrations including X chromosome aberrations
GM1734	3	EEMEM	20U 0140	40	M		46,XY,-15,+der(9),t(9;15)(9pter>9q12::15q11>15qter)mat	U	A PROBAND	257	1	
*GM3314	2	EEMEM	20U 0225	48	F	WH	46,XX/46,XX,t(5;15)(5pter>5q22::15q26>15qter;15pter>15q26::5q22>5qter)	B	PROBAND	603	2	Gardner syndrome; 40% 46,XX; 14% of cells are polyploid at P5; see GM3315A Lymphoid
*GM3255	3	EEMEM	20U 0204	3MO	M	WH	46,XY,-15,+der(15),t(8;15)(15pter>15q26::8p21>8pter)mat	U	A PROBAND	609	1	Partial trisomy 8(p); growth retardation; dysmorphic features; skeletal abnormalities; congenital heart defect

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 15													
*GM3256	3	EEMEM	20U	0204	25	F	WH 46,XX,t(8;15)(8qter>8p21::15q26>15qter; 15pter>15q26::8p21>8pter)	B	A	MOTHER	609	2	Clinically normal; balanced carrier
CHROMOSOME 16													
*GM1396	3	EEMEM	20U	0095	3	M	WH 46,XY,-10,+der(10),t(10;16)(10pter> 10q26::16q22>16qter)pat	U	A	PROBAND			Mental retardation; no speech and questionable hearing; clinically normal father's leukocytes show 46,XY,t(10;16)(q26;q22)
GM2044	2	EEMEM	20U	0164	7	M	45,XY,-16,t(14;16)(16;18)(14qter>14p12:: 16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			Slight macrocephaly; generalized hypotonia; mongoloid slant to eyes
GM2860	10	EEMEM	20U	0036	42	M	WH 46,XY,t(3;16)(3pter>3q132::16q13>16qter; 16pter>16q13::3q132>3qter)	B	A	PROBAND			Family history of recurrent abortions
*GM3005	8	EEMEM	20U	0268	28	F	WH 46,XX,t(11;16)(11pter>11q13::16p11> 16pter;16qter>16p11::11q13>11qter)	B	A	PROBAND			Phenotypically normal; mother and daughter have same balanced translocation
GM3884	7	EEMEM	10I	0354	14	F	WH 46,X,t(X;16)(Xpter>Xq26::16q24>16qter; 16pter>16q24::Xq26>Xqter)	B	A	PROBAND			Klippel-Feil syndrome; congenital heart murmur; bilaterally enlarged kidneys; mild hearing loss; inguinal hernias; normal X is late replicating; slow growing culture
GM7147	7	EEMEM	20I	0489	31	F	46,XX,t(13;16)(13pter>13q12.1::16p13.11> 16pter;16qter>16p13.11::13q12.1>13qter)	B	A	PROBAND			Clinically normal
GM2325	4	EEMEM	20U	0182	11DA	F	WH 47,XX,+der(22),t(16;22)(22pter>22q12:: 16p13>16pter)mat	U	A	PROBAND	179	1	Daughter of GM2324 Lymphoid; anophthalmia, congenital heart defect, incomplete simian crease, asymmetric ear position, hypertrophic gingiva, wide-spread nipples, hypoplastic nails, frontal bossing, & flat nasal bridge
CHROMOSOME 17													
*AG1951	12	EEMEM	20U	0201	40	M	WH 46,XY,t(17;20)(17pter>17q25::20q131> 20qter;20pter>20q131::17q25>17qter)	B		PROBAND			Xeroderma pigmentosum patient from USSR; complementation group undetermined; line LHC638
*GM0119	5	EEMEM	20U	0087	31	F	WH 46,XX,t(17;22)(17qter>17p13::22q11> 22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND			Clinically normal

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAIMS	REMARKS
							TRANSLOCATION						
CHROMOSOME 17													
*GM0271	3	EEMEM	20U 0058	28	F	WH	46,XX,t(17;19)(17pter>17q23::19p13>19pter;19qter>19p13::17q23>17qter)	B		A PARENT			Normal phenotype; 1 spontaneous abortion & 1 stillborn with anomalies
*GM1139	4	EEMEM	20U 0095	21	F	WH	46,XX,t(15;17)(15pter>15q15::17p13>17pter;17qter>17p13::15q15>15qter),t(6;13)(p21;q34)	B		A PARENT			Culture is mosaic: at passage 6, 64% carry only the t(15;17) & 36% carry both; mother of a severely defective child with the unbalanced translocation
*GM2598	3	EEMEM	20U 0109	18	F	WH	46,X,t(Y;17)(Ypter>Yq11::17p13>17pter;17qter>17p13::Yq11>Yqter)	B		A PROBAND			Gonadoblastoma; streak gonad; amenorrhea; skin fibroblast culture
*GM2808	4	EEMEM	20U 0140	30	F	WH	46,XX,t(3;17)(3qter>3p21::17p13>17pter;17qter>17p13::3p21>3pter)	B		A PROBAND			Multiple spontaneous abortions
*GM2836	4	EEMEM	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B		A PROBAND			Clinically normal; 61% of cells do not have the insertion (3;1)
GM2963	3	EEMEM	20U 0247	3	M	BL	46,XY,t(3;17)(3qter>3p21::17q25>17qter;17pter>17q25::3p21>3pter)	B		A PROBAND			Unusual facies; hirsute; broad hands & feet; teeth overgrown with fibrous tissue
GM5409	3	EEMEM	20U 0016	28	M	WH	46,XY,t(5;17)(5pter>5q33::17p12>17pter;17qter>17p12::5q33>5qter)	B		A PARENT			Clinically normal; daughter with unbalanced translocation was multiply malformed
GM6097A	3	EEMEM	20U 0417	15MO	F	WH	46,XX,-17,+der(17),t(7;17)(17qter>17p13.2::7p22.3>7pter)pat	U		A PROBAND			Lissencephaly syndrome (Miller-Dieker syndrome); typical facies; ventricular-septal defect
GM7150	5	EEMEM	20I 0489	18FW	F		47,XX,+der(13),t(13;17)(13pter>13q22::17q25>17qter)mat	U		A PROBAND			
GM7151	7	EEMEM	20I 0489	2WK	F	WH	46,X,t(X;17)(Xpter>Xq11.2::17q11.2>17qter;17pter>17q11.2::Xq11.2>Xqter)	B		A PROBAND			Multiple congenital defects including cleft palate, coloboma, Tetralogy of Fallot, and abnormal genitalia and extremities
*GM0217	6	EEMEM	20U 0143	9DA	M	WH	46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)pat	U		A PROBAND	142	1	Culture grows slowly; multiple anomalies

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 17													
*GM0216	4	EEMEM	20U	0143	29	M	WH 46,XY,t(10;17)(10pter>10q24::17p13>17pter;17qter>17p13::10q24>10qter)	B	A	FATHER	142	2	Clinically normal; balanced carrier
*GM0959	6	EEMEM	20U	0143	11	M	WH 46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)mat	U	A	MAT 3RD COU	142	3	Multiple congenital anomalies; growth & mental retardation; increased soluble glutamic oxaloacetic transaminase activity
*GM1663	13	EEMEM	20U	0113	14	M	WH 46,XY,-17,+der(17),t(13;17)(17qter>17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13; severe mental and motor retardation
*GM1555	5	EEMEM	20U	0113	13	M	WH 47,XY,+der(13),t(13;17)(13pter>13q14::17p13>17pter)mat	U	A	BRO	145	2	Mental retardation; minor dysmorphia
*GM0201	10	EEMEM	20U	0030	13	F	WH 47,XX,+21,t(1;17)(1qter>1p32::17p13>17pter;17qter>17p13::1p32>1pter)	U	A	PROBAND	337	1	Down's syndrome phenotype
GM3119	4	EEMEM	20U	0172	28	F	WH 46,XX,t(9;17)(9qter>9q22::17p11>17pter;17qter>17p11::9p22>9pter)	B	A	MOTHER	449	2	Fetus is GM3100 Amniotic; history of several spontaneous abortions
*GM3196	3	EEMEM	20U	0318	31	F	WH 46,XX,t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter),inv(5)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
*GM0988	10	EEMEM	10U	0082	3WK	M	WH 46,XY,t(9;17)(9qter>9p13::17q25>17qter;17pter>17q25::9p13>9pter)	B	A	PROBAND	702	1	Multiple congenital anomalies; 13% of cells are polyploid
CHROMOSOME 18													
*GM0017	4	EEMEM	20U	0012	30MO	M	BL 45,XY,-15,-18,+der(18),t(15;18)(18pter>18q23::15q13>15qter)	U	A	PROBAND			Multiple congenital anomalies & mental retardation
*GM0610	7	EEMEM	20U	0075	11WK	F	WH 46,XX,t(6;18)(6pter>6q21::18p11>18pter;18qter>18p11::6q21>6qter)	B	A	PROBAND			Phenotypically normal
*GM0657	3	EEMEM	20U	0107	4WK	M	WH 46,XY,del(7)(q34q36),t(7;18)(7pter>7q34::18q12.2>18qter;18pter>18q12.2::7q36>7qter)mat	U	A	PROBAND			Cleft palate; microcephaly; developmental delay; similarly affected cousin; clinically normal mother has balanced t(7;18) in lymphocytes
*GM1224	7	EEMEM	20U	0077	3MO	F	WH 46,XX,-13,+der(13),t(13;18)(13pter>13q34::18q11>18qter)pat	U	A	PROBAND			Multiple congenital anomalies

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 18													
GM1892	5	EEMEM	20U 0132	37	F	WH	46,XX,t(9;18)(9qter>9p24::18q12> 18qter;18pter>18q12::9p24>9pter)	B	A	PROBAND			
GM2044	2	EEMEM	20U 0164	7	M		45,XY,-16,t(14;16)(16;18)(14qter>14p12:: 16p11>16pter;18qter>18p1::16q12>16qter)	U	A	PROBAND			Slight macrocephaly; generalized hypotonia; mongoloid slant to eyes
*GM4125	5	EEMEM	20U 0340	1DA	M	WH	46,XY,t(15;18)(15pter>15q15::18p113> 18pter;18qter>18p113::15q15>15qter)	B	A	PROBAND			Phenotypically normal; died 4 hrs after birth
*GM5832	4	EEMEM	20U 0425	20FW	M	WH	46,XY,-18,+der(18),t(13;18)(18pter> 18q23::13q14.1>13qter)	U	A	PROBAND			Partial trisomy 13; multiple external & internal malformations including brachycephaly, bilateral epicanthus, malformed ears, hexadactyly, vitium cordis, & cystic kidneys
GM3089	6	EEMEM	20U 0299	3WK	F	WH	46,XX,-18,+der(18),t(13;18)(18oter> 18q23::13q31>13qter)pat	U	A	PROBAND	301	1	Anomalies consistent with both trisomy 13 & 18
GM5382	8	EEMEM	20U 0134	27	F	WH	46,XX,t(7;18)(7pter>7q36::18q12.2> 18qter;18pter>18q12.2::7q36>7qter)	B	A	PARENT	851	2	Clinically normal; proband not in Repository; unbalanced child showed mental retardation, developmental delay, seizures, dysmorphic facial features, syndactyly, and optic atrophy
CHROMOSOME 19													
*GM0089	5	EEMEM	20U 0043		F	WH	46,X,t(X;19)(Xpter>Xq22::19q133>19qter; 19pter>19q133::Xq22>Xqter)	B	A	PROBAND			Normal X is late replicating
*GM0271	3	EEMEM	20U 0058	28	F	WH	46,XX,t(17;19)(17pter>17q23::19p13> 19pter;19qter>19p13::17q23>17qter)	B	A	PARENT			Normal phenotype; 1 spontaneous abortion & 1 stillborn with anomalies
*GM4188	3	EEMEM	20U 0367	77	F	WH	46,XX/46,XX,t(6;19)(6pter>6q13::19p133> 19pter;19qter>19p133::6q13>6qter)	B		SPOUSE	682	2	Clinically normal; 78% of cells are 46,XX; see GM4189 Lymphoid; husband (GM4219) has Huntington's Disease
CHROMOSOME 20													
*AG1951	12	EEMEM	20U 0201	40	M	WH	46,XY,t(17;20)(17pter>17q25::20q131> 20qter;20pter>20q131::17q25>17qter)	B		PROBAND			Xeroderma pigmentosum patient from USSR; complementation group undetermined; line LHC63a

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 20													
GM1225	10	EEMEM	20U 0077	3MO	F	IN	46,XX,t(2;20)(2qter>2p21::20p13>20pter; 20qter>20p13::2p21>2pter)	B	A	PROBAND			Asiatic Indian; de novo; has multiple congenital anomalies
*GM2944A	4	EEMEM	20U 0107	2WK	M	WH	46,XY,-22,+der(20),t(20;22)(20pter> 20q11.2::22q11.2>22qter)	U	A	PROBAND			DiGeorge Syndrome
GM3876	4	EEMEM	20U 0066	33	M	WH	46,XY,t(1;20)(1qter>1p13::20q13> 20qter;20pter>20q13::1p13>1pter)	B	A	PROBAND			Phenotypically normal
*GM7792	2	EEMEM	20U 0150	14	F	WH	46,X,t(X;20)(Xpter>cen>20qter;20pter> cen>Xqter)	B	A	PROBAND			Mental retardation, seizures, & dysmorphic features; parents both have normal karyotypes; normal X is late replicating
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter> 14p11.2::20p11.1>20pter)mat,inv(9)(pter> p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	Retardation and multiple anomalies
*GM0982	3	EEMEM	20U 0148		F	BL	46,XX,t(14;20)(14qter>14p11.2::20p11.1> 20pter;20qter>20p11.1::14p11.2>14pter), inv(9)(p12q12)	B	A	MOTHER	146	2	Phenotypically normal; balanced carrier
GM5133	3	EEMEM	20U 0148	FW	M	BL	46,XY,-14,+der(14),t(14;20)(14qter> 14p11.2::20p11.1>20pter)mat	U	A	AFF BRO	146	3	Abortus
CHROMOSOME 21													
*GM0098	5	EEMEM	20U 0119	10MO	M		46,XY,-21,+der(21),t(4;21)(21qter> 21p12::4p11>4pter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM0144	5	EEMEM	20U 0012	34	F	WH	46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A	PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture; Down's syndrome; 3 male sibs have balanced translocation
GM1564	9	EEMEM	20U 0151	28	F	WH	45,XX,-21,t(1;21)(1pter>1q44::21q11> 21qter)	U	A	PROBAND			Clinically normal
GM1665	3	EEMEM	20U 0140	28	F	WH	46,XX,t(12;21)(12qter>12p11::21p11> 21pter;21qter>21p11::12p11>12pter)	B	A	PROBAND			Clinically normal
GM1700	3	EEMEM	20U 0095	6	M		46,XY,t(21;22)(21qter>21p12::22q11.2> 22qter;22pter>22q11.2::21p12>21pter)	B	A	PROBAND			Mental retardation
GM1813	3	EEMEM	20U 0066	19	F	WH	46,X,t(1;21)(1pter>1q12::21q22>21qter; 21pter>21q22::1q12>1qter)pat,t(X;15) (p11q13)	B	A	PROBAND			Menstrual dysfunction; t(X;15) present in 100% of cells at passage 6, but not seen in peripheral blood leukocytes

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 21													
*GM1881	7	EEMEM	20U	0140	3	M	WH 46,XY,t(1;21)(1qter>1p31::21q22>21qter; 21pter>21q22::1p31>1pter)	B	A	PROBAND			Sanfilippo A; deficient heparan N-sulfatase; normal Alpha-N-acetylglucosaminidase activity
GM2058	5	EEMEM	20U	0165	11DA	F	WH 46,XX/46,XX,-21,+t(21;21)(21qter>cen>21qter)	U	A	PROBAND			87%/13% at passage 8; Down's syndrome phenotype; both parents have normal karyotypes
GM3268	2	EEMEM	20U	0122	10MO	M	BL 46,XY,-21,+t(9;21)(9pter>cen>21qter)	U	A	PROBAND			Clinodactyly; short neck; bulbous nose; low set ears
GM3417	5	EEMEM	20U	0324	15	F	WH 45,XX,t(14;21)(14qter>cen>21qter)/ 46,XX,-14,+t(14;21)(14qter>cen>21qter)	U	A	PROBAND			Down's syndrome mosaic; 32% of cells are balanced 45,XX,t(14;21)
GM4610		EEMEM	20U	0080		F	46,XX,dic(14;21)(14qter>14p13::21p13> 21qter),+8/47,XX,+8	U	A	PROBAND			Additional t(14;21) present in only 25% of cells at passage 6; passage 3 at IMR
GM4862	2	EEMEM	20U	0315	24	F	WH 46,XX,t(7;21)(7pter>7q36::21q21> 21qter;21pter>21q21::7q36>7qter)	B	A	PROBAND			Clinically normal
*GM7457	4	EEMEM	20I	0504	24	F	WH 46,XX,t(1;21)(1pter>1q12::21q22.3> 21qter;21pter>21q22.3::1q12>1qter)	B	A	PROBAND			Healthy; has had two consecutive miscarriages at the 12th week of gestation
*GM0692	5	EEMEM	20U	0001	8	M	WH 45,XY,-2,-21,+der(2),t(2;21)(2pter> 2q37::21q2105>21qter)mat	U	A	PROBAND	133	1	Mentally retarded
*GM0693	5	EEMEM	20U	0001	27	F	WH 46,XX,t(2;21)(2pter>2q37::21q2105> 21qter;21pter>21q2105::2q37>2qter)	B	A	MOTHER	133	2	Two Down's phenotype daughters; one mentally retarded son; and one normal child
*GM1413	8	EEMEM	20U	0132	18MO	F	BL 46,XX,-10,+der(10),t(10;21)(10pter> 10q26::21q2105>21qter)mat	U	A	PROBAND	143	1	Down's syndrome
*GM1580	6	EEMEM	20U	0132	48	F	BL 46,XX,t(10;21)(10pter>10q26::21q21> 21qter;21pter>21q21::10q26>10qter)	B	A	MAT GRMR	143	2	Normal phenotype
*GM1399	5	EEMEM	20U	0132	20	M	BL 47,XY,+der(21),t(10;21)(21pter>21q2105:: 10q26>10qter)mat	U	A	MAT 1ST COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM0983	3	EEMEM	20U	0095	14MO	M	WH 46,XY,-21,+der(21),t(10;21)(21pter> 21q22::10q24>10qter)mat	U	A	PROBAND	144	1	Normal soluble glutamic oxaloacetic transaminase activity

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 21													
*GM0984B	8	EEMEM 20U	0095	34	F	WH	46,XX,t(10;21)(10pter>10q24::21q22>21qter;21pter>21q22::10q24>10qter)	B	A	MOTHER	144	2	Balanced carrier
*GM1730	11	EEMEM 20U	0132	5	F	BL	46,XX,-21,+der(21),t(X;21)(21qter>21p11::Xq11>Xqter)mat	U	A	PROBAND	148	1	Mental retardation and multiple anomalies; one normal X and the translocated X are late replicating
*GM0260	6	EEMEM 20U	0095	11	M	WH	46,XY/47,XY,+21/47,XY,+t(21;21)(qter>cen>qter)	U	A	PROBAND	327	1	66%/24%/10% Mosaic; formerly GM0069
GM2524	7	EEMEM 20U	0148	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21)(pter>q21::p12>qter)	U	A	PROBAND	822	1	82% of cells are 47,XY,+21 at passage 10; small head, high arched palate, flat occiput, upslanting palpebral fissures, bilateral epicanthal folds, hypotonic fingers, & developmental delay
GM5655	2	EEMEM 20U	0204	1MO	M	WH	46,XY,-21,+der(21),t(6;21)(21qter>21p11.2::6q22.2>6qter)mat	U	A	PROBAND	858	1	Multiple congenital anomalies and dysmorphic features
GM5656	2	EEMEM 20U	0204	26	F	WH	46,XX,t(6;21)(6pter>6q22.2::21p11.2>21pter;21qter>21p11.2::6q22.2>6qter)	B	A	MOTHER	858	2	Clinically normal; balanced carrier
GM6944	3	EEMEM 20U	0074	24	F	WH	46,XX,t(21;22)(21qter>21p12::22q11.1>22qter;22pter>22q11.1::21p12>21pter)	B	A	PARENT	977	1	Son has unbalanced translocation and Down's Syndrome; see GM6943 Lymphoid
CHROMOSOME 22													
*GM0005	5	EEMEM 20U	0111	30	F	WH	45,XX,t(14;22)(14qter>cen>22qter)	U	A	PROBAND			Clinically normal
*GM0084A	8	EEMEM 20U	0095	1MO	M	WH	47,XY,+der(22),t(11;22)(22pter>22q11::11q23>11qter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM0119	5	EEMEM 20U	0087	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND			Clinically normal
*GM0392	9	EEMEM 20U	0002	1	F	WH	45,XX,t(13;22)(13qter>cen>22qter)	U	A	PROBAND			Myelomonocytic leukemia
*GM0627	3	EEMEM 20U	0058	30	F	BL	46,XX,t(13;22)(13pter>13q22::22q13>22qter;22pter>22q13::13q22>13qter)	B	A	PARENT			Clinically normal; son has multiple anomalies and unbalanced translocation
*GM0980	2	EEMEM 20U	0012	33	F	WH	45,XX,-11,-22,+der(11),t(11;22)(11pter>11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION					
CHROMOSOME 22												
GM1700	3	EEMEM	20U	0095	6	M	46,XY,t(21;22)(21qter>21p12::22q11.2> 22qter;22pter>22q11.2::21p12>21pter)	B	A PROBAND			Mental retardation
*GM2944A	4	EEMEM	20U	0107	2WK	M WH	46,XY,-22,+der(20),t(20;22)(20pter> 20q11.2::22q11.2>22qter)	U	A PROBAND			DiGeorge Syndrome
GM3118	2	EEMEM	20U	0320		F	46,XX,del(11)(p13p11)/46,XX,del(11) (p13p11),t(7;22)(p15;p12)	U	A PROBAND			Bilateral Wilms tumor and bilateral aniridia; 40% of cells do not show the translocation
*GM3577	2	EEMEM	20U	0107	8MO	M WH	45,XY,-22,t(3;22)(3pter>3q29::22q11> 22qter)	U	A PROBAND			DiGeorge syndrome; absent thymus and parathyroid glands; cleft lip and palate; congenital heart disease
GM3847	8	EEMEM	15U	0353	38	F WH	46,XX,t(11;22)(11pter>11q25::22q13> 22qter;22pter>22q13::11q25>11qter)mat	B	A PROBAND			Clinically normal; mother has same translocation; neither shows any myeloproliferative disorder
GM4514	8	EEMEM	20U	0265	4	F WH	45,X/45,X,-22,+dic(X;22)(Xpter>Xq22:: 22p12>22qter)	U	A PROBAND			Frontal bossings; telecanthus; intermittent exotropia; broad nasal bridge; high arched palate; presacral dimple; hyperconvex nails; 4% 45,X; dic(X;22) is late replicating
*GM4628	3	EEMEM	20U	0383		F	46,X,t(X;22)(Xpter>Xq12::22p11>22pter; 22qter>22p11::Xq12>Xqter)	B	A PROBAND			Normal X is late replicating
*GM5401	3	EEMEM	20U	0417	3MO	M WH	45,XY,-4,-22,+der(4),t(4;22)(4pter> 4q35::22q11>22qter)mat	U	A PROBAND			Hispanic; partial DiGeorge syndrome; truncus arteriosis, type I; T-cell dysfunction; micrognathia and high arched palate; positive family history
GM7332	2	EEMEM	20U	0012	10A	M WH	46,XY,t(11;22)(11pter>11q23::22q11.2> 22qter;22pter>22q11.2::11q23>11qter)mat	B	A PROBAND			Placenta biopsy; mother has same balanced translocation
GM2325	4	EEMEM	20U	0182	11DA	F WH	47,XX,+der(22),t(16;22)(22pter>22q12:: 16p13>16pter)mat	U	A PROBAND	179	1	Daughter of GM2324 Lymphoid; anophthalmia, congenital heart defect, incomplete simian crease, asymmetric ear position, hypertrophic gingiva, wide-spread nipples, hypoplastic nails, frontal bossing, & flat nasal bridge

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMNS	REMARKS
							TRANSLOCATION						
CHROMOSOME 22													
*GM3196	3	EEMEM	20U 0318	31	F	WH	46,XX,t(17;22)(17qter>17p13::22q11> 22qter;22pter>22q11::17p13>17pter), inv(5)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
*GM3371	2	EEMEM	20U 0322	5WK	F	BL	47,XX, +der(22),t(11;22)(22pter>22q11:: 11q23>11qter)mat, inv(9)(p13q21)pat	U	A	PROBAND	613	1	Partial trisomy 22; developmental delay
GM3372	2	EEMEM	20U 0322	28	F	BL	46,XX,t(11;22)(11pter>11q23::22q11> 22qter;22pter>22q11::11q23>11qter)	B	A	MOTHER	613	2	Clinically normal
*GM4370A	3	EEMEM	20U 0007	4	F	WH	47,XX,+der(22),t(11;22)(22pter>22q11:: 11q23>11qter)mat	U	A	PROBAND	718	1	Microcephaly; earpits; micrognathia; mentally retarded
GM4403	4	EEMEM	20U 0007	30	F	WH	46,XX,t(11;22)(11pter>11q23::22q11> 22qter;22pter>22q11::11q23>11qter)	B	A	MOTHER	718	2	Clinically normal
GM6526	2	EEMEM	20U 0107	4WK	M	BL	45,XY,-22,t(13;22)(13pter>13q34::22q11.2 >22qter)	U	A	PROBAND	959	1	Skin biopsy; unusual facies: small jaw, flat philtrum, downturned upper lip, cupped ears, & sloping forehead with low hairline; unusually long fingers with distal phalanges angled upwards
GM6944	3	EEMEM	20U 0074	24	F	WH	46,XX,t(21;22)(21qter>21p12::22q11.1> 22qter;22pter>22q11.1::21p12>21pter)	B	A	PARENT	977	1	Son has unbalanced translocation and Down's Syndrome; see GM6943 Lymphoid
							RECOMBINANT						
CHROMOSOME 01													
*GM2946	8	EEMEM	20U 0265	3MO	F	WH	46,XX,rec(1),dup q,inv ins(1)(pter>p22:: q32>q31::p22>qter)pat	U	A	PROBAND			Partial trisomy 1 with multiple congenital malformations (high arched palate, hypertelorism, shield chest, earlobe anomalies, craniosynostosis, & ventricular septal defect)
CHROMOSOME 03													
GM3039	4	EEMEM	20U 0134	10MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25:: q21>qter)pat	U	A	PROBAND			Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U 0224	17MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25:: q21>qter)pat	U	A	PROBAND			Multiple congenital anomalies
*GM1253	3	EEMEM	20U 0106	4MO	M	BL	46,XY,rec(3),dup q,inv(3)(qter>p25:: q25>qter)mat	U	A	PROBAND	149	1	Parents are GM1250 & GM1252 Fibroblast; maternal half-brother is GM1251 Fibroblast; multiple congenital anomalies, patent urachus, malrotation of midgut with annular pancreas, heart murmur, cryptorchidism, & bilateral extra digit

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							RECOMBINANT					
CHROMOSOME 03												
*GM0366	3	EEMEM 20U	0003	4	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21> qter)	U	A PROBAND	560	1	Multiple anomalies; mother has inv(3)(p25q21)
CHROMOSOME 13												
*GM1570	2	EEMEM 20U	0119	16FW	M		46,XY,rec(13)dup p,inv(13)(pter>q22:: p11>pter)pat	U	A PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p11q22); severe hydrocephaly, edema, low set malformed ears, absent thumbs, & bilateral talipes
							RING CHROMOSOME					
CHROMOSOME X												
GM6563	4	EEMEM 20I	0456	11	F	WH	45,X/46,X,r(X)(p22.3>q24)	U	A PROBAND			No stigmata of Turner syndrome or hypothyroidism; pos fam hx of short stature and pubertal delay; height below the 3rd percentile; brachydactyly; no suggestion of chondrodysplasia; occ. hyperactivity; sleep disturb.; easily upset; 28% r(X)
CHROMOSOME 06												
GM0109	7	EEMEM 20U	0134	2	F	WH	46,XX,r(6)	U	A PROBAND			12% r(6); 5% 46,XX; and 83% polyploid
GM5754	2	EEMEM 20U	0204	4DA	M	WH	45,XY,-6/46,XY,r(6)	U	A PROBAND			Multiple congenital anomalies; dysmorphic features: hydrocepnaly, anal atresia, and congenital heart defect; 58% of cells show r(6) at passage 5
CHROMOSOME 09												
*GM0166	4	EEMEM 20U	0107	6	M	WH	45,XY,-9/46,XY,r(9)	U	A PROBAND	2002	1	44% of cells have r(9); 56% are 45,XY,-9; trigonocephaly, hypertelorism, systolic murmur, & multiple facial malformations
CHROMOSOME 13												
GM0285	3	EEMEM 20U	0107	4DA	M	BL	46,XY,r(13)	B	A PROBAND			Multiple congenital anomalies
*GM0729	8	EEMEM 20U	0142	14	M	WH	45,XY,-13/46,XY,r(13)	U	A PROBAND			Mental retardation; 76% of cells have r(13); ring is unstable; slow growing culture

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
RING CHROMOSOME													
CHROMOSOME 13													
GM3321	2	EEMEM	20U 0066	11DA	F	WH	46,XX,r(13)	U	A	PROBAND			Greek; bilateral cleft lip & palate; microcephaly, microphthalmos; hypertelorism; 16% of cells have a fragment & no r(13)
GM5563	3	R1640	20U 0345	FW	M	WH	46,XY,r(13)	U	A	PROBAND			Breakpoint may be q14; severely malformed fetus; anencephaly, imperforato anus, urethral meatus, severe talipes, syndactyly, horseshoe cataracts, bilateral retinal dysplasia, & type IV polycystic kidneys
GM6799	2	EEMEM	20U 0107	6MO	F	WH	46,XX,r(13)/46,XX,i(13)(qter>cen>qter)	U	A	PROBAND			84% of cells are 46,XX,i(13) at P5; cleft lip & palate; scalp hemangioma; microphthalmia
CHROMOSOME 14													
GM3003	6	EEMEM	20U 0134	6	M	WH	46,XY,r(14)	U	A	PROBAND			Growth retardation; hypotonia; seizures
GM3233	2	EEMEM	20U 0122	1WK	M	WH	45,XY,-14/46,XY,r(14)	U	A	PROBAND			Congenital malformations; 88% of cells are 46,XY,r(14) at passage 4
GM7364	9	EEMEM	20U 0354	5	M	WH	46,XY,r(14)(p12>q32.33)	U	A	PROBAND			Mental retardation; small; thin; limited speech; mild micrognathia & camptodactyly; brachydactyly; hypoplastic scrotum; scoliosis; absent right kidney; 2 fused ribs; seizures; 92% of cells show r(14) at P10
CHROMOSOME 17													
GM6047	2	EEMEM	20U 0417	4MO	M	WH	46,XY,r(17)(p13.3q25.3)	U	A	PROBAND			Lissencephaly syndrome (Miller-Dieker syndrome)
CHROMOSOME 18													
GM1118	8	EEMEM	20U 0150	30MO	F	WH	45,XX,-18/46,XX,r(18)	U	A	PROBAND			78% of cells have r(18) at passage 18; multiple congenital anomalies
CHROMOSOME 21													
GM6137	3	EEMEM	20U 0369	NB	M	BL	46,XY,r(21)(p13>q22)	U	A	PROBAND	924	1	Fibroblast culture (umbilical cord); incarcerated inguinal hernia; no apparent dysmorphic features; same patient as GM6199

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMB	FAMN	REMARKS
							RING CHROMOSOME						
CHROMOSOME 21													
GM6199	9	EEMEM	20U 0369	NB	M	BL	46,XY,r(21)(p13>q22)	U	A	PROBAND	924	1	Fibroblast culture (foreskin); incarcerated inguinal hernia; no apparent dysmorphic features; same patient as GM6137; 94% of cells have r(21) at P7
CHROMOSOME 22													
GM6420	2	EEMEM	20U 0326	2	M	BL	46,XY,r(22)	U	A	PROBAND			Severe mental retardation; small head, slight epicanthal folds, small chin, elfin ears, & only 2 creases on hands; normal urine amino & organic acids; both parents have normal karyotypes
							MARKER CHROMOSOME						
CHROMOSOME X													
*GM4024A	7	EEMEM	20U 0061	29	M	BL	46,fra(X)(q27),Y 4% of cells show fra(X) with FudR	B	A	PROBAND	588	1	X-linked mental retardation; seizures; macro-orchidism; 52% of leukocytes show fra(X)(q27); see GM4025B Lymphoid
*GM4026	2	EEMEM	20U 0061	35	M	WH	46,fra(X)(q27),Y 2% of cells show fra(X)	B	A	PROBAND	589	1	X-linked mental retardation; macro-orchidism; positive family history
GM5131	6	EEMEM	20U 0148	3	M	WH	46,fra(X)(q27),Y 8% of cells show fra(X) with FudR	B	A	PROBAND	819	1	Fragile X-linked mental retardation; similarly affected brother and uncle
GM5848A	5	EEMEM	20U 0427	4	M	WH	46,fra(X)(q27),Y 12% of cells show fra(X) with FudR	B	A	PROBAND	872	1	Moderate mental retardation; large ears and long face with prognathic appearance; undefined connective tissue dysplasia; 50% of lymphocytes show fra(X)
GM5847	6	EEMEM	20U 0427	36	F	WH	46,X,fra(X)(q27) 4% of cells show fra(X) with FudR	B	A	MOTHER	872	2	Heterozygote for X-linked mental retardation; clinically normal but dull; mild connective tissue dysplasia; one affected son; fra(X)(q27) present in 40% of lymphocytes
CHROMOSOME 10													
*GM5425A	14	EEMEM	20U 0418	28	F	WH	46,XX,fra(10)(q25) fra(10)(q25) in 12% of cells at P17	B	A	PROBAND	855	1	Short stature, late menarche, irregular menses, small uterus & ovaries, aortic insufficiency, myopia, & cataracts; see GM5523A Lymphoid

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMD	FAMM	REMARKS
							INVERSION						
CHROMOSOME 01													
GM0803	3	EEMEM	20U	0002	2	M	WH	46,XY,del(1)(pter>q21.3::q23>qter),inv(1)(pter>p13::q12>p13::q12>qter)	U	A	PROBAND		Unusual facies
GM3126	2	EEMEM	20U	0148	4	F	BL	46,XX,del inv(1)(pter>p13.1::q21.1>p13.1::q21.1>q42.3::)	U	A	PROBAND		Multiple congenital abnormalities; Tetralogy of Fallot
CHROMOSOME 02													
*GM0735	17	EEMEM	20U	0077	17	F	WH	46,X,dic(X)(qter>p11::p11>qter),inv(2)(pter>p11::q13>p11::q13>qter)	U	A	PROBAND		Turner's syndrome
*GM2365	4	EEMEM	20U	0213	14	M	WH	46,XY,inv(2)(pter>p11.2::q13>p11.2::q13>qter)	B	A	PROBAND		Coffin-Lowry syndrome; mental retardation, short stature, unusual facies, and tapering fingers; positive family history
CHROMOSOME 03													
GM3039	4	EEMEM	20U	0134	10MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND		Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U	0224	17MO	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND		Multiple congenital anomalies
*GM1253	3	EEMEM	20U	0106	4MO	M	BL	46,XY,rec(3),dup q,inv(3)(qter>p25::q25>qter)mat	U	A	PROBAND	149 1	Parents are GM1250 & GM1252 Fibroblast; maternal half-brother is GM1251 Fibroblast; multiple congenital anomalies, patent urachus, malrotation of midgut with annular pancreas, heart murmur, cryptorchidism, & bilateral extra digit
*GM1252	3	EEMEM	20U	0106	22	F	BL	46,XX,inv(3)(pter>p25::q25>p25::q25>qter),inv(9)(pter>p11::q1>p11::q1>qter)	B	A	MOTHER	149 2	Phenotypically normal; sons are GM1251 and GM1253 Fibroblast
AG3594		EEMEM	20U	0044	30	M	WH	46,XY,inv(3)(q23q29),t(3;13)(p21;q32),inv(9)(p11q34)	B		AFF BRO	546 2	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3593A; parents are first cousins
*GM0366	3	EEMEM	20U	0003	4	M	WH	46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)	U	A	PROBAND	560 1	Multiple anomalies; mother has inv(3)(p25q21)
CHROMOSOME 04													
GM0413	4	EEMEM	20U	0056	1	M		46,XY,inv(4)(pter>p14::q2>p14::q2>qter)mat	B	A	PROBAND	150 1	Minor malformations

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INVERSION						
CHROMOSOME 04													
GM0414	3	EEMEM	20U 0056	24	F		46,XX,inv(4)(pter>p14::q2>p14::q2>qter)	B	A	MOTHER	150	2	Clinically normal; balanced carrier
CHROMOSOME 05													
GM3883	9	EEMEM	15U 0224	3	F	WH	46,XX,inv(5)(pter>q15::q33>q15::q33>qter)pat	B	A	PROBAND			Developmental delay; hypertelorism; brushfield spots; flat nasal bridge
GM2335	3	EEMEM	20U 0125	21	M	WH	46,XY,inv(5)(pter>p1505::q12>p1505::q12>qter)	B	A	PROBAND	217	1	G6PD(B)+ Xg(a) antigen absent; hypogonadism; parents are GM2334 & GM2336 Fibroblast
*GM2137	2	EEMEM	20U 0012	2	F	WH	47,XX,inv(5)(pter>p13::q13>p13::q13>qter)mat, +mar	U	A	PROBAND	453	1	Marker may be rec(5)(p1q1)mat; multiple congenital anomalies; mental retardation
*GM3196	3	EEMEM	20U 0318	31	F	WH	46,XX,inv(5)(pter>p13::q13>p13::q13>qter),t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND	518	1	See GM3197 Lymphoid; no hematological disorder; habitual aborter; normal phenotype
CHROMOSOME 06													
GM2957	4	EEMEM	20U 0315	23	F	WH	46,XX,inv(6)(pter>p23::q21>p23::q21>qter)	B	A	PARENT			A child has partial trisomy 6p+
GM2206	3	EEMEM	20U 0147	17	F	WH	46,XX,inv(6)(pter>p24::p21>p24::p21>qter)pat	B	A	PROBAND	208	1	Unclassified Ehlers-Danlos syndrome variant; formerly GM1514; hydroxylysine deficient; not lysyl hydroxylase deficient
GM2205	4	EEMEM	20U 0147	51	M	WH	46,XY,inv(6)(pter>p24::p21>p24::p21>qter)	B	A	FATHER	208	2	Clinically normal; formerly GM1515
CHROMOSOME 07													
GM3666	4	EEMEM	20U 0326	34	M		47,XY,+dic(15)(pter>q11::q11>pter)/47,XY,+dic(15)(pter>q11::q11>pter),inv(7)(pter>p22::q32>p22::q32>qter)	U	A	PROBAND			Clinically normal; inv(7) is present in 38% of cells
CHROMOSOME 09													
GM0425	12	R1640	20U 0013	10FW	M		47,XY,+8,inv(9)(pter>p11::q21>p11::q21>qter)	U	A	PROBAND			Spontaneous abortion
GM6256		EEMEM	20U 0452	7MO	F	WH	46,XX,inv(9)(pter>p11::q21>p11::q21>qter)pat	B		PROBAND			Zellweger syndrome; delayed development; hepatomegaly; rotary nystagmus; large anterior fontanelle; protuberant eyes; questionable retinitis pigmentosa; increased C26:C22 fatty acid ratio; passage 3 at IMR

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INVERSION						
CHROMOSOME 09													
*GM0981	3	EEMEM	20U 0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter>14p11.2::20p11.1>20pter)mat,inv(9)(pter>p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	Retardation and multiple anomalies
*GM0982	3	EEMEM	20U 0148		F	BL	46,XX,t(14;20)(14qter>14p11.2::20p11.1>20pter;20qter>20p11.1::14p11.2>14pter),inv(9)(p12q12)	B	A	MOTHER	146	2	Phenotypically normal; balanced carrier
*GM1252	3	EEMEM	20U 0106	22	F	BL	46,XX,inv(3)(pter>p25::q25>p25::q25>qter),inv(9)(pter>p11::q1>p11::q1>qter)	B	A	MOTHER	149	2	Phenotypically normal; sons are GM1251 and GM1253 Fibroblast
*GM1251	2	EEMEM	20U 0106	4	M	BL	46,XY,inv(9)(pter>p11::q1>p11::q1>qter)mat	B	A	MAT HALF BRO	149	4	Clinically normal; mother is GM1252; maternal half-brother is GM1253
*GM0447	7	EEMEM	20U 0030	1	M	WH	46,XY,inv(9)(pter>p11::q13>p11::q13>qter)pat	B	A	PROBAND	151	1	Congenital malformations
*GM0445	7	EEMEM	20U 0030	26	M	WH	46,XY,inv(9)(pter>p11::q13>p11::q13>qter)	B	A	FATHER	151	2	Clinically normal
*GM0450	7	EEMEM	20U 0030	33	M	WH	46,XY,inv(9)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	152	1	Clinically normal; skin biopsy; same pt. as GM0451
*GM0451	7	EEMEM	20U 0030	33	M	WH	46,XY,inv(9)(pter>p13::q13>p13::q13>qter)	B	A	PROBAND	152	1	Same patient as GM0450; testicular tissue fibroblast culture
*GM1920	2	EEMEM	20U 0061	23	M	WH	47,XY,+21,inv(9)(pter>p11::q13>p11::q13>qter)mat,t(8;14)(8pter>8q11::14q11>14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	Down's syndrome phenotype; see GM1921A Lymphoid
GM3120	5	EEMEM	20U 0172	5	F		45,X,inv(9)(p11q13)/46,XY,inv(9)(p11q13)	U	A	PROBAND	450	1	Cultures from left gonad (GM3121) and right gonad (GM3122) also available; 82% of cells are 45,X,inv(9); slow growing culture
AG3594		EEMEM	20U 0044	30	M	WH	46,XY,inv(3)(q23q29),t(3;13)(p21;q32),inv(9)(p11q34)	B		AFF BRO	546	2	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3593A; parents are first cousins
GM3900	3	EEMEM	20U 0358	15	F	BL	46,XX,inv(9)(pter>p11::q13>p11::q13>qter)	B		PROBAND	582	1	Abdominal tumor biopsy; Gardner syndrome; similarly affected father; 14% of cells are polyploid at P8; same patient as GM3901

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INVERSION						
CHROMOSOME 09													
GM3901	3	EEMEM	20U	0358	15	F	BL 46,XX,inv(9)(pter>p11::q13>p11::q13>qter)	B		PROBAND	582	1	Skin biopsy; Gardner syndrome; similarly affected father; 11% of cells are polyploid at P5; same patient as GM3900
*GM3371	2	EEMEM	20U	0322	5WK	F	BL 47,XX, +der(22),t(11;22)(22pter>22q11::11q23>11qter)mat, inv(9)(p13q21)pat	U	A	PROBAND	613	1	Partial trisomy 22; developmental delay
GM5882	10	EEMEM	20U	0078	25MO	F	WH 49,XXXXX,inv(9)(pter>p22::q11>p22::q11>qter)/48,XXXX,inv(9)(pter>p22::q11>p22::q11>qter)	U	A	PROBAND	914	1	Developmental delay; microcephaly; small for age; low set ears; bilateral epicanthal folds; decreased muscle tone; low total finger-ridge count; parents both have normal karyotypes; see GM6061A Lymphoid; 52% 49,XXXXX,inv(9); 16% polypld.
CHROMOSOME 10													
GM6109	4	EEMEM	20U	0224	18FW	F	46,XX,inv(10)(pter>p11.2::q21>p11.2::q21>qter)	B	A	PROBAND			
*GM0448	7	EEMEM	20U	0030	1	M	WH 46,XY,inv(10)(pter>p11::q21>p11::q21>qter)pat	B	A	PROBAND	153	1	Congenital malformations
*GM0452	7	EEMEM	20U	0030	22	M	WH 46,XY,inv(10)(pter>p11::q21>p11::q21>qter)	B	A	FATHER	153	2	Clinically normal
CHROMOSOME 13													
*GM0437	3	EEMEM	20U	0066	32	F	WH 46,XX,inv(13)(qter>q21::p1>q21)	B	A	PROBAND			
*GM1570	2	EEMEM	20U	0119	16FW	M	46,XY,rec(13)dup p,inv(13)(pter>q22::p11>pter)pat	U	A	PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p11q22); severe hydrocephaly, edema, low set malformed ears, absent thumbs, & bilateral talipes
							INSERTION						
CHROMOSOME 01													
*GM2836	4	EEMEM	20U	0263	43	M	WH 46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B	A	PROBAND			Clinically normal; 61% of cells do not have the insertion (3;1)
*GM2946	8	EEMEM	20U	0265	3MO	F	WH 46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)pat	U	A	PROBAND			Partial trisomy 1 with multiple congenital malformations (high arched palate, hypertelorism, shield chest, earlobe anomalies, craniosynostosis, & ventricular septal defect)

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INSERTION						
CHROMOSOME 01													
GM0860	11	EEMEM	20U 0036	13	F	WH	46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly
GM0861	7	EEMEM	20U 0036	40	M	WH	46,XY,ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter;1pter>1q25::1q32>1qter)	B	A	FATHER	128	2	Balanced carrier
CHROMOSOME 03													
GM2486	3	EEMEM	20U 0140	5MO	F		46,XX,ins(3)(pter>q21::q21>q27::q21>qter)	U	A	PROBAND			Both parents are diabetic and have normal karyotypes
*GM2836	4	EEMEM	20U 0263	43	M	WH	46,XY,t(9;17)(q12;p11)/46,XY,t(9;17)(q12;p11),ins(3;1)(q21;q25q44)	B	A	PROBAND			Clinically normal; 61% of cells do not have the insertion (3;1)
CHROMOSOME 05													
GM0860	11	EEMEM	20U 0036	13	F	WH	46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25::1q32)::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly
GM0861	7	EEMEM	20U 0036	40	M	WH	46,XY,ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter;1pter>1q25::1q32>1qter)	B	A	FATHER	128	2	Balanced carrier
*GM1221A	9	EEMEM	20U 0021	18MO	M	BL	46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	PROBAND	138	1	Partial trisomy of 6q15q27; peculiar facies; short & stubby fingers; bilateral clinodactyly of 5th finger; bilateral simian creases but left is atypical
*GM1222	5	EEMEM	20U 0021	23	F	BL	46,XX,ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter;6pter>6q15::6q27>6qter)	B	A	MOTHER	138	2	Mother and half brother have same balanced insertion; normal phenotype
*GM1524	3	EEMEM	20U 0021	19	M	BL	46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	COU	138	3	Growth and mental retardation; peculiar facies
CHROMOSOME 06													
*GM1221A	9	EEMEM	20U 0021	18MO	M	BL	46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	PROBAND	138	1	Partial trisomy of 6q15q27; peculiar facies; short & stubby fingers; bilateral clinodactyly of 5th finger; bilateral simian creases but left is atypical
*GM1222	5	EEMEM	20U 0021	23	F	BL	46,XX,ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter;6pter>6q15::6q27>6qter)	B	A	MOTHER	138	2	Mother and half brother have same balanced insertion; normal phenotype
*GM1524	3	EEMEM	20U 0021	19	M	BL	46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	COU	138	3	Growth and mental retardation; peculiar facies

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INSERTION						
CHROMOSOME 10													
GM5296	2	EEMEM	20U 0204	1MO	F	WH	46,XX,-11,+der(11)inv ins(11;10)(11pter> 11p13::10q21>10q24::11p13>11qter)	U	A	PROBAND	845	1	Multiple congenital malformations; dysmorphic features; intrauterine growth retardation; heart murmur; cleft palate; equinovarus deformity; microcephaly; coloboma of rt. iris; clinodactyly; reduced RBC catalase activity
GM5297	4	EEMEM	20U 0204	21	F	WH	46,XX,inv ins(11;10)(11pter>11p13::10q21>10q24::11p13>11qter;10pter>10q21::10q24>10qter)	B	A	MOTHER	845	2	Phenotypically normal; mother and grandfather carry the same translocation; normal red blood cell catalase activity
CHROMOSOME 11													
GM5296	2	EEMEM	20U 0204	1MO	F	WH	46,XX,-11,+der(11)inv ins(11;10)(11pter> 11p13::10q21>10q24::11p13>11qter)	U	A	PROBAND	845	1	Multiple congenital malformations; dysmorphic features; intrauterine growth retardation; heart murmur; cleft palate; equinovarus deformity; microcephaly; coloboma of rt. iris; clinodactyly; reduced RBC catalase activity
GM5297	4	EEMEM	20U 0204	21	F	WH	46,XX,inv ins(11;10)(11pter>11p13::10q21>10q24::11p13>11qter;10pter>10q21::10q24>10qter)	B	A	MOTHER	845	2	Phenotypically normal; mother and grandfather carry the same translocation; normal red blood cell catalase activity
							ISOCHROMOSOME						
CHROMOSOME X													
*GM0088	5	EEMEM	20U 0012	19	F	WH	46,X,i(X)(qter>cen>qter)	U	A	PROBAND			Buccal smear was 18% sex chromatin positive; Turner's syndrome
GM2595	6	EEMEM	20U 0073	33	F	WH	46,X,i(X)(qter>cen>qter)	U	A	PROBAND			Turner's syndrome
CHROMOSOME Y													
*GM1709	4	EEMEM	20U 0109	11	F	WH	46,X,i(Y)(qter>cen>qter)	U	A	PROBAND			Gonadal dysgenesis; 8 of 12 karyotypes have an additional t(1;5); biopsy of gonadal streak
GM3774	2	EEMEM	20U 0315	16	F	WH	45,X,+frag/46,X,i(Y)(qter>cen>qter)	U	A	PROBAND			Turner's syndrome mosaic; 72% of cells lack the i(Y)
CHROMOSOME 13													
GM6799	2	EEMEM	20U 0107	6MO	F	WH	46,XX,r(13)/46,XX,i(13)(qter>cen>qter)	U	A	PROBAND			84% of cells are 46,XX,i(13) at P5; cleft lip & palate; scalp hemangioma; microphthalmia

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMD	FAMM	REMARKS
							ISOCHROMOSOME						
CHROMOSOME 13													
GM6873A		EEMEM	20U	0107		F	WH 46,XX,i(13)(qter>cen>qter)	U	A	PROBAND			Cleft lip & palate; empty right eye socket; radial denervation of wrists; duplication of great toes; scalp hemangioma; passage 3 at IMR; 46,XX,r(13) was also seen in blood lymphocytes
							DUPLICATION						
CHROMOSOME 01													
*GM2946	8	EEMEM	20U	0265	3MO	F	WH 46,XX,rec(1),dup q,inv ins(1)(pter>p22::q32>q31::p22>qter)pat	U	A	PROBAND			Partial trisomy 1 with multiple congenital malformations (high arched palate, hypertelorism, shield chest, earlobe anomalies, craniosynostosis, & ventricular septal defect)
CHROMOSOME 02													
*GM2848	3	EEMEM	20U	0070	4	M	WH 46,XY,-2,+dup(2)(2qter>2q33::2p25>2qter)	U	A	PROBAND			In lymphocytes, 28% of the cells have r(2)(2p25>2q33) instead of the duplication; mild mental retardation, unusual facies, bilateral single transverse palmar creases, & systolic heart murmur
CHROMOSOME 03													
GM3039	4	EEMEM	20U	0134	10MO	M	WH 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Gross failure to thrive; multiple congenital anomalies
GM3085	9	EEMEM	20U	0224	17MO	M	WH 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)pat	U	A	PROBAND			Multiple congenital anomalies
*GM1253	3	EEMEM	20U	0106	4MO	M	BL 46,XY,rec(3),dup q,inv(3)(qter>p25::q25>qter)mat	U	A	PROBAND	149	1	Parents are GM1250 & GM1252 Fibroblast; maternal half-brother is GM1251 Fibroblast; multiple congenital anomalies, patent urachus, malrotation of midgut with annular pancreas, heart murmur, cryptorchidism, & bilateral extra digit
*GM0366	3	EEMEM	20U	0003	4	M	WH 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter)	U	A	PROBAND	560	1	Multiple anomalies; mother has inv(3)(p25q21)
CHROMOSOME 06													
*AG3593A		EEMEM	20U	0044	28	M	WH 46,XY,t(5;8)(q11;q22),dup(6)(p21>p25),9qh+	B		PROBAND	546	1	Werner's syndrome; passage 4 at IMR; similarly affected brother is AG3594; parents are first cousins

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DUPLICATION						
CHROMOSOME 09													
*GM6074	5	EEMEM	20U	0003	2	F	WH 46,XX,rec(9)dup(9)inv ins(9)(pter> q22.1::q34.3>q34.1::q22.1>qter)mat	U	A	PROBAND			Phenotype suggestive of Marfan syndrome but no ocular or cardiac anomalies
CHROMOSOME 13													
*GM1570	2	EEMEM	20U	0119	16FW	M	46,XY,rec(13)dup p,inv(13)(pter>q22::p11>pter)pat	U	A	PROBAND	481	1	Paternal karyotype is 46,XY,inv(13)(p11q22); severe hydrocephaly, edema, low set malformed ears, absent thumbs, & bilateral talipes
							DICENTRIC CHROMOSOME						
CHROMOSOME X													
*GM0314	16	EEMEM	20U	0030	60	F	WH 45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A	PROBAND			Ovarian dysgenesis without Turner stigmata; 70% of cells have the dicentric chromosome; dicentric X is late replicating
GM0339	3	EEMEM	20U	0030	25	F	WH 45,X/46,X,dic(X)(qter>p11::p11>qter)	U	A	PROBAND			60%/40% Mosaic; 83% of 45,X cells have additional t(2;4); a subline with two dic(X) is present in 4% of cells; Turner's syndrome
*GM0735	17	EEMEM	20U	0077	17	F	WH 46,X,dic(X)(qter>p11::p11>qter),inv(2)(pter>p11::q13>p11::q13>qter)	U	A	PROBAND			Turner's syndrome
GM3935	8	EEMEM	20U	0342	20	F	WH 46,X,dic(X)(pter>q27::q27>pter)	U	A	PROBAND			Primary amenorrhea, underdeveloped uterus, bilateral streak ovaries; dic(X) is late replicating
GM4430A	9	HF10	20I	0078	31	F	WH 45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A	PROBAND			Amenorrhea; polycystic ovaries; short stature; 70% 45,X at passage 13
GM4514	8	EEMEM	20U	0265	4	F	WH 45,X/45,X,-22,+dic(X;22)(Xpter>Xq22::22p12>22qter)	U	A	PROBAND			Frontal bossings; telecanthus; intermittent exotropia; broad nasal bridge; high arched palate; presacral dimple; hyperconvex nails; 4% 45,X; dic(X;22) is late replicating
GM5396	2	EEMEM	20U	0416	2MO	F	WH 45,X,dic(X;22)(Xqter>Xp11::22p12>22qter)	U	A	PROBAND			Pixie-like appearance; simple ears; high palatine arch; micrognathia; hypotonia; poor weight gain; X chromosome portion of derivative chromosome is late replicating

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DICENTRIC CHROMOSOME						
CHROMOSOME X													
GM6960	4	EEMEM	20U 0326	26	F	BL	46,X,dic(X)(pter>q22::q22>pter)	U	A	PROBAND			Atrophic vagina and narrow, small uterus
GM7149	7	EEMEM	20I 0489	2WK	F	WH	45,X,dic(X;22)(Xqter>Xp22.1::22p11.2>22qter)	U	A	PROBAND			Congenital heart failure, large ductus arteriosis; minor congenital anomalies
GM7213	3	EEMEM	20U 0390	14	F	BL	46,X,dic(X)(pter>q28::q28>pter)	U	A	PROBAND			Culture established from ovarian biopsy; small infantile uterus and cervix; undergoing chemotherapy for previous malignancy; dic(X) is late labelling
CHROMOSOME 13													
*GM3518		EEMEM	20U 0265	4MO	F	BL	46,XX,dic(13)(13pter>13q22::13q11>13pter)	U	A	PROBAND			Severe microcephaly; oligodactyly; coloboma; midline scalp defect; hypoplastic nose; passage 2 at IMR
GM3786	6	EEMEM	20U 0353	19	F	WH	45,XX,dic(13;14)(13qter>13p11::14p11>14qter)	U	A	PROBAND			Greek; clinically normal
CHROMOSOME 14													
GM3786	6	EEMEM	20U 0353	19	F	WH	45,XX,dic(13;14)(13qter>13p11::14p11>14qter)	U	A	PROBAND			Greek; clinically normal
CHROMOSOME 15													
GM3666	4	EEMEM	20U 0326	34	M		47,XY,+dic(15)(pter>q11::q11>pter)/47,XY,+dic(15)(pter>q11::q11>pter),inv(7)(pter>p22::q32>p22::q32>qter)	U	A	PROBAND			Clinically normal; inv(7) is present in 38% of cells
CHROMOSOME 21													
GM2524	7	EEMEM	20U 0148	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21)(pter>q21::p12>qter)	U	A	PROBAND	822	1	82% of cells are 47,XY,+21 at passage 10; small head, high arched palate, flat occiput, upslanting palpebral fissures, bilateral epicanthal folds, hypotonic fingers, & developmental delay
CHROMOSOME 22													
GM5396	2	EEMEM	20U 0416	2MO	F	WH	45,X,dic(X;22)(Xqter>Xp11::22p12>22qter)	U	A	PROBAND			Pixie-like appearance; simple ears; high palatine arch; micrognathia; hypotonia; poor weight gain; X chromosome portion of derivative chromosome is late replicating
GM7149	7	EEMEM	20I 0489	2WK	F	WH	45,X,dic(X;22)(Xqter>Xp22.1::22p11.2>22qter)	U	A	PROBAND			Congenital heart failure, large ductus arteriosis; minor congenital anomalies

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME					
CHROMOSOME X												
*GM7503	12	EEMEM	20U 0506	10	F	WH	46,X,+der(X),t(X;2)(Xpter>Xq28::2p21>2pter)mat	U	A PROBAND			Congenital heart disease; growth & mental retardation; alacrima; unusual triangular facies; strabismus; frontal bossing; simple, low set, posteriorly rotated ears; hypotonia
CHROMOSOME 01												
GM1229	3	EEMEM	20U 0007	3	F	BL	46,XX,-1,+der(1),t(1;2)(1qter>1p36::2q31>2qter)pat	U	A PROBAND	129	1	Multiple congenital anomalies & mental retardation
GM3369	3	EEMEM	20U 0322	23	F	WH	46,XX,-1,+der(1),t(1;10)(1pter>1q44::10p11>10pter)mat	U	A PROBAND	612	1	Developmental delay; similarly affected sister
CHROMOSOME 02												
*GM0501	3	EEMEM	20U 0014	5	M	WH	46,XY,-2,+der(2),t(2;4)(2qter>2p25::4q21>4qter)pat	U	A PROBAND	131	1	Growth and mental retardation
*GM4371	3	EEMEM	20U 0354	4	M	OT	46,XY,-2,+der(2),t(2;5)(2qter>2p25::5q33>5qter)mat	U	A PROBAND	715	1	Black/Caucasian; multiple anomalies; partial trisomy 5q
CHROMOSOME 04												
*GM5336	2	EEMEM	20U 0107	14DA	M	WH	46,XY,-4,+der(4),t(3;4)(4qter>4p15::3p21.3>3pter)mat	U	A PROBAND			"Greek helmet face"; shallow orbits & protruding eyes; congenital heart disease; hypospadias; undescended testes
GM5394	6	EEMEM	20U 0326	3MO	M	WH	46,XY,-4,+der(4),t(4;9)(4pter>4q35::9p21>9pter)mat	U	A PROBAND			Prominent forehead, low set ears, flat nasal bridge, high arched palate, micrognathia, short neck, bilateral simian creases, and hypoplastic toenails
*GM5401	3	EEMEM	20U 0417	3MO	M	WH	45,XY,-4,-22,+der(4),t(4;22)(4pter>4q35::22q11>22qter)mat	U	A PROBAND			Hispanic; partial DiGeorge syndrome; truncus arteriosis, type I; T-cell dysfunction; micrognathia and high arched palate; positive family history
*GM1220	3	EEMEM	20U 0066	2	F		46,XX,-4,+der(4),t(4;7)(4qter>4p16::7q34>7qter)mat	U	A PROBAND	136	1	Hypotonia, clubbed feet, and cardiac abnormality with cyanotic spells
*GM1183	4	EEMEM	20U 0007	NB	M	WH	46,XY,-4,+der(4),t(4;12)(4qter>4p1608::12p11>12pter)pat	U	A PROBAND	709	2	Small for dates; abnormal facies; coloboma; multiple anomalies

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 05													
*GM0071	15	EEMEM	20U	0015	19	F	WH 46,XX,-5,der(5),t(5;?)(p1;?)mat	U	A	PROBAND			Cri du Chat; deletion assumed to be derived rather than a simple deletion; ATCC CCL 90
*GM0344	4	EEMEM	20U	0066	7	M	WH 45,XY,-5,-15,+der(5),t(5;15)(5qter>5p14::15q11>15qter),t(9;11)(9qter>9p24::11q23>11qter;11pter>11q23::9p24>9pter)	U	A	PROBAND			Cri du Chat
GM0860	11	EEMEM	20U	0036	13	F	WH 46,XX,-5,+der(5),ins(5;1)(5pter>5q15::1q25>1q32::5q15>5qter)pat	U	A	PROBAND	128	1	Culture grows slowly
GM3493	2	EEMEM	20U	0204	5MO	F	WH 46,XX,-5,+der(5),t(5;6)(5pter>5q35::6p22>6pter)pat	U	A	PROBAND	135	1	Partial trisomy 6p; psychomotor retardation; multiple congenital anomalies
*GM1524	3	EEMEM	20U	0021	19	M	BL 46,XY,-5,+der(5),ins(5;6)(5pter>5q33::6q15>6q27::5q33>5qter)mat	U	A	COU	138	3	Growth and mental retardation; peculiar facies
CHROMOSOME 06													
*GM0144	5	EEMEM	20U	0012	34	F	WH 46,XX,-6,+der(6),t(6;21)(6qter>6p25::21q11>21qter)mat	U	A	PROBAND			13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture; Down's syndrome; 3 male sibs have balanced translocation
*GM3647	2	EEMEM	20U	0204	41	M	WH 46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	PROBAND	297	1	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1; similarly affected half-brother
*GM3648	2	EEMEM	20U	0204	32	M	WH 46,XY,-6,+der(6),t(1;6)(6qter>6p25::1q32>1qter)mat	U	A	HALF BRO	297	2	Growth and mental retardation; mildly dysmorphic; partial trisomy of the distal long arm of chromosome 1
CHROMOSOME 07													
GM7216	5	EEMEM	20U	0224	25FW	F	WH 46,XX,-7,+der(7),t(3;7)(7pter>7q36::3p21.3>3pter)	U	A	PROBAND			Cyclops; holoprosencephaly; proboscis; both parents have normal karyotypes
CHROMOSOME 09													
*GM3563	2	EEMEM	20U	0148	1MO	M	WH 46,XY,-9,+der(9),t(3;9)(9qter>9p24::3q12>3qter)mat	U	A	PROBAND			Multiple congenital anomalies

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMD	FAMN	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 09													
*GM1414	5	EEMEM	20U	0068	28	F	BL 46,X,-X,+der(9),t(X;9)(9pter>9q34::Xq13> Xqter)mat	U	A	PROBAND	140	1	Turner's syndrome; in blood leukocytes and fibroblasts abnormal X is late replicating
GM1734	3	EEMEM	20U	0140	40	M	46,XY,-15,+der(9),t(9;15)(9pter>9q12::15q11>15qter)mat	U	A	PROBAND	257	1	
*GM1387	4	EEMEM	20U	0061	14	F	WH 46,XX,-13,+der(9),t(9;13)(9pter>9q22::13q12>13qter)mat or 46,XX,-13,+der(9),t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A	PROBAND	369	1	Result of Adj-2 segregation; see also GM1388 Lymphoid; profoundly retarded; clinical features of trisomy 9p syndrome
GM5347	2	EEMEM	20U	0074	2MO	M	WH 46,XY,-9,+der(9),t(1;9)(9qter>9p24::1q32>1qter)mat	U	A	PROBAND	849	1	Partial trisomy 1q; cup-shaped ear, small flattened nose, deep philtrum, downward slanting eyes, club feet, pyloric stenosis, VSD, and hyperconvex nails; mother is GM5348 Lymphoid
*GM5319	4	EEMEM	20U	0107	24FW	F	WH 47,XX,+der(9),t(9;13)(9pter>9q31.2::13q32.1>13qter)mat	U	A	PROBAND	1042	1	Mother with balanced translocation is GM7456 Lymphoid
CHROMOSOME 10													
*GM1396	3	EEMEM	20U	0095	3	M	WH 46,XY,-10,+der(10),t(10;16)(10pter>10q26::16q22>16qter)pat	U	A	PROBAND			Mental retardation; no speech and questionable hearing; clinically normal father's leukocytes show 46,XY,t(10;16)(q26;q22)
GM3241A	3	EEMEM	20U	0016	10MO	F	WH 45,XX,-10,-13,+der(10),t(10;13)(10pter>10q26::13q12>13qter)	U	A	PROBAND			Multiple congenital anomalies; both parents have normal karyotypes
*GM3689	6	EEMEM	20U	0247	24	F	WH 46,XX,-10,+der(10),t(8;10)(10qter>10p15::8p21.1>8pter)mat	U	A	PROBAND			Mental & growth retardation; delayed sexual maturation; multiple congenital anomalies
GM4993	4	EEMEM	20U	0148	1MO	F	WH 46,XX,-10,+der(10),t(4;10)(10pter>10q26::4q31>4qter)mat	U	A	PROBAND			Congenital heart disease, genito-urinary anomalies, malrotation of bowel, & dysmorphic features
GM1848	3	EEMEM	20U	0179	35	M	WH 46,XY,-10,+der(10),t(2;10)(10pter>10q26::2p23>2pter)mat	U	A	MAT 1ST COU	134	2	Possible red cell acid phosphatase variant

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 10													
GM1649	4	EEMEM 20U	0179	26	M	WH	46,XY,-10,+der(10),t(2;10)(10pter> 10q26::2p23>2pter)mat	U	A	MAT 1ST COU	134	3	Possible red cell acid phosphatase variant; slow growing culture
*GM1413	8	EEMEM 20U	0132	18MO	F	BL	46,XX,-10,+der(10),t(10;21)(10pter> 10q26::21q2105>21qter)mat	U	A	PROBAND	143	1	Down's syndrome
CHROMOSOME 11													
*GM0980	2	EEMEM 20U	0012	33	F	WH	45,XX,-11,-22,+der(11),t(11;22)(11pter> 11q25::22q11>22qter)	U	A	PROBAND			69% of cells also show t(1;4)(q25;p12)
GM5296	2	EEMEM 20U	0204	1MO	F	WH	46,XX,-11,+der(11)inv ins(11;10)(11pter> 11p13::10q21>10q24::11p13>11qter)	U	A	PROBAND	845	1	Multiple congenital malformations; dysmorphic features; intrauterine growth retardation; heart murmur; cleft palate; equinovarus deformity; microcephaly; coloboma of rt. iris; clinodactyly; reduced RBC catalase activity
CHROMOSOME 12													
*GM4127	4	EEMEM 20U	0346	6MO	F	BL	46,XX,-12,+der(12),t(3;12)(12qter> 12p13::3p21>3pter)	U	A	PROBAND			Partial trisomy 3p; microcephaly; brachycephalic; cleft lip & palate
GM1535	6	EEMEM 20U	0002	16	F	BL	46,XX,-12,+der(12),t(5;12)(12pter> 12q24::5q33>5qter)mat	U	A	PROBAND	139	1	
GM2819	3	EEMEM 20U	0107	7MO	F	BL	46,XX,-12,+der(12),t(9;12)(12pter> 12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2820 Lymphoid
*GM6033	1	EEMEM 20U	0107	5	F	BL	46,XX,-12,+der(12),t(9;12)(12pter> 12q24.31::9p13>9pter)mat	U	A	PROBAND	998	1	Congenital heart disease, loose joints, and unusual facies
CHROMOSOME 13													
*GM1224	7	EEMEM 20U	0077	3MO	F	WH	46,XX,-13,+der(13),t(13;18)(13pter> 13q34::18q11>18qter)pat	U	A	PROBAND			Multiple congenital anomalies
GM7150	5	EEMEM 20I	0489	18FW	F		47,XX,+der(13),t(13;17)(13pter>13q22:: 17q25>17qter)mat	U	A	PROBAND			
*GM1555	5	EEMEM 20U	0113	13	M	WH	47,XY,+der(13),t(13;17)(13pter>13q14:: 17p13>17pter)mat	U	A	BRO	145	2	Mental retardation; minor dysmorphia
CHROMOSOME 14													
*GM1750	2	EEMEM 20U	0070	30MO	M	WH	47,XY,+der(14),t(9;14)(14pter>14q21:: 9p24>9pter)mat	U	A	PROBAND	141	1	Three first cousins have the same partial trisomy 14; growth retardation, facial dysplasia, & developmental delay

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 14													
*GM0981	3	EEMEM 20U	0148	4	F	BL	46,XX,-14,+der(14),t(14;20)(14qter>14p11.2::20p11.1>20pter)mat,inv(9)(pter>p13::q13>p13::q13>qter)	U	A	PROBAND	146	1	Retardation and multiple anomalies
GM5133	3	EEMEM 20U	0148	FW	M	BL	46,XY,-14,+der(14),t(14;20)(14qter>14p11.2::20p11.1>20pter)mat	U	A	AFF BRO	146	3	Abortus
*GM0074	10	EEMEM 10U	0111	23	N	WH	47,Y,t(X;14)(Xpter>Xq13::14q32>14qter;14pter>14q32::Xq13>Xqter),+der(14),t(X;14)(14pter>14q32::Xq13>Xqter)mat	U	A	PROBAND	147	1	Klinefelter's syndrome; der(14) is late replicating in 88% of cells; trisomic for 14pter>14q32
GM3462	2	EEMEM 20U	0204	3MO	F	WH	47,XX,+der(14),t(9;14)(14pter>14q13::9p22>9pter)mat	U	A	PROBAND	227	1	Growth retardation and congenital malformations
GM3810	4	EEMEM 10I	0354	18	F	WH	46,XX,-14,+der(14),t(4;14)(14qter>14p12::4p15>4pter)mat	U	A	PROBAND	552	2	Partial trisomy 4p; mental and developmental retardation
*GM4409	2	EEMEM 20U	0204	6	M	WH	46,XY,-14,+der(14),t(2;14)(14pter>14q32::2p23>2pter)mat	U	A	PROBAND	721	1	Mental retardation and congenital malformations
CHROMOSOME 15													
*GM0118	5	EEMEM 20U	0052	29	F	WH	46,XX,-15,+der(15),t(Y;15)(15qter>15p1::Yq11>Yqter)pat	U	A	PROBAND			Uterine tissue fibroblast culture; phenotypically normal
*GM2488		EEMEM 20U	0140	23	M	WH	46,XY,-15,+der(15),t(5;15)(15qter>15p11::5p13>5pter)mat	U	A	PROBAND	225	1	Passage 6 at IMR
*GM3255	3	EEMEM 20U	0204	3MO	M	WH	46,XY,-15,+der(15),t(8;15)(15pter>15q26::8p21>8pter)mat	U	A	PROBAND	609	1	Partial trisomy 8(p); growth retardation; dysmorphic features; skeletal abnormalities; congenital heart defect
CHROMOSOME 17													
GM6097A	3	EEMEM 20U	0417	15MO	F	WH	46,XX,-17,+der(17),t(7;17)(17qter>17p13.2::7p22.3>7pter)pat	U	A	PROBAND			Lissencephaly syndrome (Miller-Dieker syndrome); typical facies; ventricular-septal defect
*GM0217	6	EEMEM 20U	0143	9DA	M	WH	46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)pat	U	A	PROBAND	142	1	Culture grows slowly; multiple anomalies
*GM0959	6	EEMEM 20U	0143	11	M	WH	46,XY,-17,+der(17),t(10;17)(17qter>17p13::10q24>10qter)mat	U	A	MAT 3RD COU	142	3	Multiple congenital anomalies; growth & mental retardation; increased soluble glutamic oxaloacetic transaminase activity

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME						
CHROMOSOME 17													
*GM1663	13	EEMEM	20U	0113	14	M	WH 46,XY,-17,+der(17),t(13;17)(17qter>17p13::13q14>13qter)mat	U	A	PROBAND	145	1	Partial trisomy 13; severe mental and motor retardation
CHROMOSOME 18													
*GM5832	4	EEMEM	20U	0425	20FW	M	WH 46,XY,-18,+der(18),t(13;18)(18pter>18q23::13q14.1>13qter)	U	A	PROBAND			Partial trisomy 13; multiple external & internal malformations including brachycephaly, bilateral epicanthus, malformed ears, hexadactyly, vitium cordis, & cystic kidneys
CHROMOSOME 20													
*GM2944A	4	EEMEM	20U	0107	2WK	M	WH 46,XY,-22,+der(20),t(20;22)(20pter>20q11.2::22q11.2>22qter)	U	A	PROBAND			DiGeorge Syndrome
CHROMOSOME 21													
*GM0098	5	EEMEM	20U	0119	10MO	M	46,XY,-21,+der(21),t(4;21)(21qter>21p12::4p11>4pter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM1399	5	EEMEM	20U	0132	20	M	BL 47,XY,+der(21),t(10;21)(21pter>21q2105::10q26>10qter)mat	U	A	MAT 1ST COU	143	3	Mental retardation and ear anomalies; 1st cousin once removed
*GM0983	3	EEMEM	20U	0095	14MO	M	WH 46,XY,-21,+der(21),t(10;21)(21pter>21q22::10q24>10qter)mat	U	A	PROBAND	144	1	Normal soluble glutamic oxaloacetic transaminase activity
*GM1730	11	EEMEM	20U	0132	5	F	BL 46,XX,-21,+der(21),t(X;21)(21qter>21p11::Xq11>Xqter)mat	U	A	PROBAND	148	1	Mental retardation and multiple anomalies; one normal X and the translocated X are late replicating
GM5655	2	EEMEM	20U	0204	1MO	M	WH 46,XY,-21,+der(21),t(6;21)(21qter>21p11.2::6q22.2>6qter)mat	U	A	PROBAND	858	1	Multiple congenital anomalies and dysmorphic features
CHROMOSOME 22													
*GM0084A	8	EEMEM	20U	0095	1MO	M	WH 47,XY,+der(22),t(11;22)(22pter>22q11::11q23>11qter)mat	U	A	PROBAND			Multiple congenital anomalies
*GM3371	2	EEMEM	20U	0322	5WK	F	BL 47,XX, +der(22),t(11;22)(22pter>22q11::11q23>11qter)mat, inv(9)(p13q21)pat	U	A	PROBAND	613	1	Partial trisomy 22; developmental delay
*GM4370A	3	EEMEM	20U	0007	4	F	WH 47,XX,+der(22),t(11;22)(22pter>22q11::11q23>11qter)mat	U	A	PROBAND	718	1	Microcephaly; earpits; micrognathia; mentally retarded

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REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAM#	REMARKS
							DELETION					
CHROMOSOME X												
GM1941	3	EEMEM	20U 0012	13	F	WH	45,X/46,X,del(X)(pter>q11:)	U	A PROBAND			44% of fibroblasts are 45,X; Turner's syndrome phenotype; slow growing culture
*GM2470	9	EEMEM	20U 0030	55	F	WH	46,X,del(X)(pter>p22.2::p11.4>qter)	U	A PROBAND			Clinically normal; no Turner stigmata; in leukocytes and fibroblasts del(X) is late replicating
*GM3624A	3	EEMEM	20U 0204	23	F	WH	46,X,del(X)(pter>q26:)	U	A PROBAND			Phenotypically normal; normal fertility; deleted X is late replicating; both parents have normal karyotypes; 2 live-born infants: 1 child with del(X) & trisomy 21 & 1 chromosomally & phenotypically normal female
*GM3923	5	R1640	15U 0003	25	F	WH	46,X,del(X)(pter>q13::q22>qter)	U	A PROBAND			English; clinically normal; one child has same chromosome abnormality; deleted X is late replicating
GM7148	10	EEMEM	20I 0489	18	F	AI	46,X,del(X)(pter>q22.3:)	U	A PROBAND			Primary amenorrhea; elevated FSH; hypogonadism
*GM3827	2	EEMEM	20U 0326	18	F	BL	46,X,del(X)(pter>q2101:)	U	A PROBAND	557	1	Primary ovarian failure; secondary amenorrhea; increased luteinizing & follicle stimulating hormone; right ovary biopsy; same patient as GM3828
GM3828	3	EEMEM	20U 0326	18	F	BL	46,X,del(X)(pter>q2101:)	U	A PROBAND	557	1	Primary ovarian failure; secondary amenorrhea; increased luteinizing & follicle stimulating hormones; left ovary biopsy; same patient as GM3827; deleted X is late replicating
CHROMOSOME Y												
GM2668	4	EEMEM	20U 0229	41	A	WH	45,X/46,X,del(Y)(pter>q12:)	U	A PROBAND			Male pseudohermaphrodite; several of Turner syndrome stigmata; mixed gonadal dysgenesis; 33% of cells are 45,X
GM2730	10	EEMEM	20U 0109	4DA	A	WH	45,X/46,X,del(Y)(pter>q11:)	U	A PROBAND			Breakpoint is above heterochromatic region; 33% of cells are 45,X; ambiguous genitalia

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME Y													
GM3595	6	EEMEM	20U 0125	11	A	WH	45,X/46,X,del(Y)(pter>q11:)	U	A	PROBAND			Mixed gonadal dysgenesis; ambiguous external genitalia; psychosomatic retardation; H-Y antigen positive; 84% have del (Y); breakpoint is probably q11
CHROMOSOME 01													
*GM0214	8	EEMEM	20U 0030	2	M	WH	46,XY,del(1)(pter>q25::q32>qter)	U	A	PROBAND			Growth hormone deficiency & hypothyroidism
GM0803	3	EEMEM	20U 0002	2	M	WH	46,XY,del(1)(pter>q21.3::q23>qter), inv(1)(pter>p13::q12>p13::q12>qter)	U	A	PROBAND			Unusual facies
*GM2025	4	EEMEM	20U 0126	30MO	M	WH	46,XY,del(1)(pter>q21::q25>qter)	U	A	PROBAND			Microcephaly; low set ears; small eyes; high arched palate with bifid uvula; abdominal hernia; dislocated hip; hypotonia; bilateral small kidneys; parents both have normal karyotypes
GM2906A	8	EEMEM	20U 0014	3WK	F	WH	46,XX,del(1)(pter>q23.3::q32.1>qter)	U	A	PROBAND			Bilateral cleft lip & palate; brachydactyly and clinodactyly
GM3126	2	EEMEM	20U 0148	4	F	BL	46,XX,del inv(1)(pter>p13.1::q21.1>p13.1::q21.1>q42.3:)	U	A	PROBAND			Multiple congenital abnormalities; Tetralogy of Fallot
*GM4940	4	EEMEM	20U 0390	22FW	F	WH	46,XX,del(1)(pter>q25::q32>qter)	U	A	PROBAND			Partial monosomy chromosome 1; polysplenism; cephalohematoma
GM5302	5	EEMEM	20U 0411	NB	F	WH	46,XX,del(1)(pter>q41::q43>qter)	U	A	PROBAND			Prominent occiput, mild hypertelorism, short neck, wrinkled skin, talipes equinovarus of feet, and cleft palate
GM6038	6	EEMEM	20I 0326	1DA	M		46,XY,del(1)(pter>q23.3::q25.3>qter)	U	A	PROBAND			Low set ears; webbed neck; cleft lip & palate; displaced hip joint; no testes; rocker bottom-left foot; club foot-right; hypoplastic lungs; horseshoe kidney; simian creases-both hands
*GM6473	8	R1640	20U 0032	3	F	WH	46,XX,del(1)(pter>q42:)	U	A	PROBAND			67% of control human acid B-glucosidase (GBA) activity; somatic cell hybrids with a single del(1) were negative for GBA by both immunoprecipitation and natural substrate enzyme assays

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REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION					
CHROMOSOME 02												
*GM0945	2	EEMEM	20U 0077	19MO	F	OR	46,XX,del(2)(qter>p24:)	U	A PROBAND			Failure to thrive; severe retardation; heterozygous BA phenotype at ACP-1 locus; Cantonese
*GM1138	3	EEMEM	20U 0095	1	M	BL	46,XY,del(2)(qter>p23:)	U	A PROBAND			Severe growth and mental retardation; multiple anomalies; heterozygous BA phenotype at ACP-1 locus
GM3125	4	EEMEM	20U 0148	7DA	M	WH	46,XY,del(2)(pter>q23::q32.2>qter)	U	A PROBAND			Multiple congenital abnormalities; slow growing culture
GM3918	5	EEMEM	20U 0204	7	F	WH	46,XX,del(2)(pter>q31::q33>qter)	U	A PROBAND			Dysmorphic features; motor retardation; superficially resembles Seckel syndrome
GM4513	7	EEMEM	20U 0056	FW	F	WH	47,XX,+2/47,XX,+del(2)(qter>p23:)	U	A PROBAND			Spontaneous abortion; 24% of cells show 47,XX,+2
CHROMOSOME 03												
GM7873	1	EEMEM	20U 0204	2WK	M	WH	46,XY,del(3)(qter>p25:)	U	A PROBAND			Intrauterine growth retardation; congenital heart disease; multiple dysmorphic features; both parents have normal karyotypes
CHROMOSOME 04												
*GM0072	15	EEMEM	20U 0015	11	M	WH	46,XY,del(4)(qter>p14:)	U	A PROBAND			Wolf's syndrome; 4p- syndrome
*GM2003	4	EEMEM	20U 0095	11	F	WH	46,XX,del(4)(pter>q33:)	U	A PROBAND			
*GM3013	3	EEMEM	20U 0148	1MO	F	WH	46,XX,del(4)(pter>q31:)	U	A PROBAND			Multiple congenital anomalies
*GM4045B	16	EEMEM	20U 0044	30MO	M	WH	46,XY,del(4)(q31.1q31.3),t(2;4)(2pter>2q14::4q31.3>4qter;4pter>4q31.1::2q14>2qter)	U	A PROBAND			Mental retardation, facial dysmorphy, & unusual dermatoglyphics; hemizygous at MN blood group locus
GM4126	12	EEMEM	20U 0364	1	M	WH	46,XY,del(4)(qter>p1503:)	U	A PROBAND			Wolf's syndrome; 4p- syndrome
GM7267	7	EEMEM	20U 0326	MO	M	WH	46,XY,del(4)(pter>q31.3:)	U	A PROBAND			

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME 04													
*GM0343	3	EEMEM	20U 0066	3	M		46,XY,del(4)(qter>p14:)	U	A	PROBAND	347	1	Wolf's syndrome; 4p- syndrome
CHROMOSOME 05													
GM2337	3	EEMEM	20U 0066	35	F	WH	46,XX,del(5)(qter>p1403:)	U	A	PROBAND			Cri du Chat
GM2800	4	EEMEM	20U 0148	3WK	F	BL	46,XX,del(5)(qter>p1405:)	U	A	PROBAND			Not Cri du Chat syndrome
GM3912	3	EEMEM	20U 0315	3	M	WH	45,X,del(5)(qter>p14:)	U	A	PROBAND			Cri du Chat syndrome; mental retardation; HY antigen positive in peripheral blood lymphocytes; normal plasma testosterone & male external genitalia; lymphocytes show same karyotype as fibroblasts; both parents have normal karyos.
CHROMOSOME 06													
GM6222	5	EEMEM	20U 0134	2MO	F	WH	46,XX,del(6)(qter>p24:)	U	A	PROBAND			Dandy-Walker cyst/hydrocephalus; Peter's anomaly of the eyes; hypertelorism; ear tags; low set ears; normal Hageman factor level in blood
CHROMOSOME 07													
*GM1059	5	EEMEM	10U 0149	50DA	F	WH	46,XX,del(7)(pter>q22::q32>qter)	U	A	PROBAND			Hypertonia, seizures, feeding difficulty, & multiple congenital anomalies
GM3240	8	EEMEM	20U 0016	7DA	M	WH	46,XY,del(7)(pter>q34:)	U	A	PROBAND			Microcephaly & other congenital anomalies
*GM3733	2	EEMEM	20U 0204	7	M	WH	46,XY,del(7)(pter>q34:)	U	A	PROBAND			Growth and psychomotor retardation; mild dysmorphic features; bilateral coloboma of retina and choroidea; normal factor XII (Hageman) level
GM7412	1	EEMEM	20U 0107	7	M	WH	46,XY,del(7)(pter>q32:)	U	A	PROBAND			Developmental delay; abnormal neurological examination; microcephaly, hypospadias, heart murmur, facial and extremity abnormalities; both parents have normal karyotypes
CHROMOSOME 08													
GM1549	8	EEMEM	20U 0140	7	M		46,XY,del(8)(qter>p21:)	U	A	PROBAND			Multiple congenital anomalies & mental retardation

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME 08													
*GM3134	2	EEMEM	20U 0204	3MO	M	WH	46,XY,del(8)(pter>q13::q22>qter)	U	A	PROBAND			Multiple congenital anomalies
GM3999	3	EEMEM	20U 0204	1MO	F	BL	46,XX,del(8)(pter>q24:)	U	A	PROBAND			Multiple congenital anomalies; respiratory failure; hypotonia; both parents have normal karyotypes
CHROMOSOME 09													
*GM0870	2	EEMEM	20U 0015	22	F	WH	46,XX,del(9)(qter>p13:)	U	A	PROBAND			Severe mental retardation & congenital anomalies
*GM1667	7	EEMEM	20U 0002	13	F	WH	46,XX,del(9)(qter>p22:)	U	A	PROBAND			
*GM1893	4	EEMEM	20U 0140	3	M		46,XY,del(9)(pter>q11::q21.3>qter)	U	A	PROBAND			Mental retardation, multiple anomalies, seizures
GM2356	2	EEMEM	20U 0140	3	M		46,XY,del(9)(9qter>9p22:)	U	A	PROBAND			Dysplasia
*GM3226	2	EEMEM	20U 0081	9MO	M	BL	47,XY,+del(9)(9pter>9q11:)mat	U	A	PROBAND	602	1	Multiple malformations & mental retardation; see GM5067 Lymphoid
GM3225	4	EEMEM	20U 0326	32	F	BL	46,XX/47,XX,+del(9)(9pter>9q11:)	U	A	MOTHER	602	2	36% 46,XX in lymphocytes, 98% 46,XX in fibroblasts; short stature; mentally slow; see GM5066 Lymphoid
CHROMOSOME 10													
*GM3047	7	EEMEM	20U 0016	5	M	WH	46,XY,del(10)(qter>p11:)	U	A	PROBAND			Psychomotor retardation, trigonocephaly, and multiple congenital anomalies
*GM5299	3	EEMEM	20U 0204	10DA	F	WH	46,XX,del(10)(pter>q26:)	U	A	PROBAND			Intrauterine growth retardation; microcephaly; dysmorphic features; congenital heart disease (truncus type I)
GM6937	3	EEMEM	20U 0418	27MO	F	WH	46,XX,del(10)(qter>p13:)	U	A	PROBAND	974	1	Developmental delay and dysmorphic features; parents both have normal karyotypes; see GM6936 Lymphoid
CHROMOSOME 11													
GM2008	6	EEMEM	20U 0069	2WK	F	BL	46,XX,del(11)(pter>q24:)	U	A	PROBAND			Multiple congenital anomalies & bilateral colobomata

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							DELETION						
CHROMOSOME 11													
GM3118	2	EEMEM	20U	0320		F	46,XX,del(11)(p13p11)/46,XX,del(11)(p13p11),t(7;22)(p15;p12)	U	A	PROBAND			Bilateral Wilms tumor and bilateral aniridia; 40% of cells do not show the translocation
GM3808	2	EEMEM	20U	0320	9	M	WH 46,XY,del(11)(pter>p13::p11>qter)	U	A	PROBAND			AGR triad; seizures; Wilms tumor
*GM2859	12	EEMEM	20U	0036	3	F	WH 46,X,del(11)(p11.2p1300),t(X;11)(Xpter>Xq11.1::11p1300>11pter;11qter>11p11.2::Xq11.1>Xqter)	U	A	PROBAND	617	1	See GM3316 Lymphoid; South American; in lymphoblast line, normal X is late replicating; mental retardation with bizarre dysmorphology syndrome: epicanthus, hypertelorism, & trigonocephaly
GM6938B	3	EEMEM	20U	0148	3WK	M	WH 46,XY,del(11)(pter>p13::p14>qter)	U	A	PROBAND	1074	1	Micropenis; cryptorchidism; bilateral aniridia; optic nerve atrophy; genital skin fibroblast culture; see GM7736 Lymphoid
CHROMOSOME 13													
*GM2718	7	EEMEM	15U	0244	7	F	46,XX,del(13)(pter>q12.3::q21.2>qter)	U	A	PROBAND			D-deletion Retinoblastoma
*GM2719	6	EEMEM	20U	0244	29	M	46,XY,del(13)(pter>q22::q32>qter)	U	A	PROBAND			Bilateral colobomas, multiple congenital anomalies of hands & feet
*GM1142A	8	EEMEM	20U	0094	27MO	F	WH 46,XX,del(13)(pter>q14.1::q22.1>qter)	U	A	PROBAND	187	1	D-deletion Retinoblastoma; bilateral; see GM1484 Lymphoid & GM3022 SV40 Transformed Fibroblast; at P6, a subline with t(3;16)(q12;q12.1) is present in 15% of cells
*GM0509A	11	EEMEM	20U	0143	16MO	F	BL 46,XX,del(13)(pter>q14:)	U	A	PROBAND	345	1	Short term lymphocytes show r(13); formerly GM0250; mental & growth retardation, bilateral cleft lip & palate, and other congenital anomalies; no Retinoblastoma; slow growing culture
GM5877	8	EEMEM	20U	0047	2	F	46,XX,del(13)(pter>q14.1::q21.2>qter)	U	A	PROBAND	1025	1	Bilateral, sporadic Retinoblastoma; negative family history; normal growth and development except for delayed speech

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	REMARKS
							DELETION			
CHROMOSOME 16										
*GM2346	2	EEMEM 20U	0204	3MO	M	WH	46,XY,del(16)(pter>q22:)	U	A PROBAND	Hydrocephalus, hypospadias, & minor dysmorphic features
GM5875A	6	EEMEM 20U	0148	7MO	F	WH	46,XX,del(16)(pter>p11.2::p11.1>qter)	U	A PROBAND	Low set ears, hypertelorism with wide Mongolian slant, beaked nose, Pierre Robin anomalies, and hypoplastic nipples
CHROMOSOME 18										
GM1727	13	EEMEM 20U	0142	45	M	WH	46,XY,del(18)(qter>p11:)	U	A PROBAND	Mentally retarded; aberrations involving chromosomes 1, 10, 15, & 17 were also found in all cells examined
GM2980	2	EEMEM 20U	0107	12	M	WH	46,XY,del(18)(pter>q2108:)	U	A PROBAND	
GM3767	2	EEMEM 20U	0204	3	M	WH	46,XY,del(18)(qter>p11:)	U	A PROBAND	Clinical features of 18p-; lymphocytes show 70% 18p- and 30% i(18q)
CHROMOSOME 21										
GM4111	2	EEMEM 20U	0368	28	F	WH	46,XX,t(4;5)(4pter>4q31.1::5p15>5pter; 5qter>5p15::4q31.1>4qter),del(21)(qter> p11:)	U	A PARENT	Clinically normal; has 1 normal child; 1 child died & appeared to have Down's phenotype
							TETRAPLOID			
CHROMOSOME 4N										
*GM6979	8	EEMEM 30I	0480	2MO	M	WH	92,XXYY	U	A PROBAND	Patient expired at two months of age; mult. congenit. anomal.: unusual facies, cleft palate, congenital heart disease, & unusual foot abnormalities; macrocytic anemia, hypersegmented neutrophils, and megaloblastic changes in the marrow
							TRIPLOID			
CHROMOSOME 3N										
*GM1322	3	EEMEM 20U	0095	2DA	A	BL	69,XXY	U	A PROBAND	Meningomyelocele; phenotypically female; G6PD type AB; both X chromosomes are active and early-replicating
*GM1672	8	EEMEM 10U	0088	1DA	M	WH	69,XXY	U	A PROBAND	G6PD type B

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRIPLOID						
CHROMOSOME 3N													
GM3591	6	EEMEM	20U 0107	26FW	♀	WH	69,XXY	U	A	PROBAND			Multiple anomalies
GM4376A	8	EEMEM	20I 0078	25FW	F	WH	69,XXX	U	A	PROBAND			Hydrocephalic; stillborn; no other abnormalities
							ANEUPLOID						
CHROMOSOME X													
*GM0157	12	EEMEM	20U 0030	28	♀	WH	49,XXXXY,t(4;11)(4pter>4q35::11q23>11qter;11pter>11q23::4q35>4qter)	U	A	PROBAND			Severe mental retardation; sexual infantilism
GM0254	8	EEMEM	20U 0075	10FW	F	BL	47,XXX	U	A	PROBAND			76% interphase nuclei have 2 sex chromosome bodies; 15% have one; no evidence for mosaicism
*GM0314	16	EEMEM	20U 0030	60	F	WH	45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A	PROBAND			Ovarian dysgenesis without Turner stigmata; 70% of cells have the dicentric chromosome; dicentric X is late replicating
GM0325	4	EEMEM	20U 0125	30	M	WH	47,XXY	U	A	PROBAND			Sardinian; heterozygous for G6PD deficiency (med. type), Xg(a) antigen present
GM0775A	8	EEMEM	20U 0107	13	F		45,X	U	A	PROBAND			Turner's syndrome
*GM0857	3	EEMEM	20U 0095	1DA	F	WH	45,X	U	A	PROBAND			Turner's syndrome
*GM1137	6	R1640	10U 0017	1MO	M	BL	48,XXY,+21,t(6;10)(6qter>6p25::10p11>10pter;10qter>10p11::6p25>6pter)	U	A	PROBAND			Down's syndrome features
GM1176	2	EEMEM	20U 0107	8	F	WH	45,X	U	A	PROBAND			Turner's syndrome
GM1441	2	EEMEM	20U 0074	3MO	M	WH	45,X	U	A	PROBAND			Tissue from right testis; lymphocytes showed 45,X/46,X,iso(Y); 80%/20%; coarctation of aorta, cardiomegaly, & Meckel's diverticulum

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID					
CHROMOSOME X												
GM1941	3	EEMEM	20U 0012	13	F	WH	45,X/46,X,del(X)(pter>q11:)	U	A PROBAND			44% of fibroblasts are 45,X; Turner's syndrome phenotype; slow growing culture
*GM2521	9	EEMEM	15U 0222	24	M	WH	48,XXYY	U	A PROBAND			Patient has high levels of gonadotrophins: FSH, LH, and testosterone; tall; thin; aggressive behavior; unusual appearance
GM3133	6	EEMEM	20U 0273	10FW	F		46,X,+15	U	A PROBAND			Culture established from a spontaneous abortus; grows slowly; 141% of normal pyruvate kinase-3 activity
GM3623	6	EEMEM	20U 0340	NB	F	BL	48,XXX,+18	U	A PROBAND			Clinical features of variant trisomy 18
GM4375A	4	EEMEM	20U 0271	21FW	M	WH	48,XXYY	U	A PROBAND			Klinefelter's syndrome variant
GM4430A	9	HF10	20I 0078	31	F	WH	45,X/46,X,dic(X)(qter>p22::p22>qter)	U	A PROBAND			Amenorrhea; polycystic ovaries; short stature; 70% 45,X at passage 13
GM4514	8	EEMEM	20U 0265	4	F	WH	45,X/45,X,-22,+dic(X;22)(Xpter>Xq22::22p12>22qter)	U	A PROBAND			Frontal bossings; telecanthus; intermittent exotropia; broad nasal bridge; high arched palate; presacral dimple; hyperconvex nails; 4% 45,X; dic(X;22) is late replicating
GM4626	2	EEMEM	20U 0144	21FW	F	WH	47,XXX	U	A PROBAND			Abortus; clinically normal phenotype
GM4965	7	EEMEM	20U 0271	1MO	M	OR	48,XXY,+21	U	A PROBAND			Down's syndrome phenotype
GM5009	5	EEMEM	20U 0397	6MO	F	WH	49,XXXXX	U	A PROBAND			Growth & developmental delay, hypotonia, mongoloid facies, & unusual hands & feet
GM6563	4	EEMEM	20I 0456	11	F	WH	45,X/46,X,r(X)(p22.3>q24)	U	A PROBAND			No stigmata of Turner syndrome or hypothyroidism; pos fam hx of short stature and pubertal delay; height below the 3rd percentile; brachydactyly; no suggestion of chondrodysplasia; occ. hyperactivity; sleep disturb.; easily upset; 28% r(X)

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAM#	REMARKS
							ANEUPLOID					
CHROMOSOME X												
*GM0326	3	EEMEM	20U	0125	6	M	WH 49,XXXXY	U	A PROBAND	164	4	Normal RBC G6PD level; Xg(a) antigen positive; severe retardation; hypogonadism; mother is GM0321 Fibroblast
*GM1415E	3	EEMEM	20U	0061	35	F	WH 48,XXXX	U	A PROBAND	370	1	Develop delay; mod retard; delayed language develop; abnorm dermatoglyphics; bilat epicanth folds; sm mandible; bilat dermato-lipomas; lens opacities; high arched palate; mildly hypotonic; normal menses; see GM1416B Lymphoid
GM1723	2	EEMEM	20U	0203	23	F	WH 45,X	U	A PROBAND	394	1	Skin biopsy; fibroblast culture from uterine tissue also available (GM1724); patient has focal endometrial hyperplasia
GM3102	5	EEMEM	20U	0172	18FW	M	WH 47,XXY	U	A PROBAND	448	1	See GM3091 Amniotic; Klinefelter's syndrome
GM0562	4	EEMEM	20U	0095	14	F	BL 45,X/46,XX	U	A PROBAND	598	1	4 cells 46,XX, 46 cells 45,X; tissue from left gonad; gonad revealed testicular rather than ovarian tissue; same pt. as GM0563
GM0563	4	EEMEM	20U	0095	14	F	BL 45,X	U	A PROBAND	598	1	Only 45,X cells found; tissue from right gonad; gonad revealed testicular rather than ovarian tissue; same pt. as GM0562
GM5882	10	EEMEM	20U	0078	25MO	F	WH 49,XXXXX,inv(9)(pter>p22::q11>p22::q11>qter)/48,XXXX,inv(9)(pter>p22::q11>p22::q11>qter)	U	A PROBAND	914	1	Developmental delay; microcephaly; small for age; low set ears; bilateral epicanthal folds; decreased muscle tone; low total finger-ridge count; parents both have normal karyotypes; see GM6061A Lymphoid; 52% 49,XXXXX,inv(9); 16% polyploid.
CHROMOSOME Y												
*GM2521	9	EEMEM	15U	0222	24	M	WH 48,XXYY	U	A PROBAND			Patient has high levels of gonadotrophins: FSH, LH, and testosterone; tall; thin; aggressive behavior; unusual appearance

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#2	REMARKS
							ANEUPLOID						
CHROMOSOME Y													
GM4375A	4	EEMEM	20U 0271	21FW	M	WH	48,XXYY	U	A	PROBAND			Klinefelter's syndrome variant
GM1250	3	EEMEM	20U 0106	23	M	BL	47,XYY	U	A	FATHER	149	3	Husband of GM1252; father of GM1253; phenotypically normal
GM0978	7	EEMEM	20U 0143	15	F	WH	45,X/46,XY	U	A	PROBAND	334	1	Formerly GM0188; 97% of cells are 45,X; tissue from right gonadal streak
CHROMOSOME 02													
*GM3576	10	EEMEM	20U 0326	13FW	M	WH	48,XY,+2,+21	U	A	PROBAND			Spontaneous abortus
GM4513	7	EEMEM	20U 0056	FW	F	WH	47,XX,+2/47,XX,+del(2)(qter>p23:)	U	A	PROBAND			Spontaneous abortion; 24% of cells show 47,XX,+2
CHROMOSOME 06													
GM5754	2	EEMEM	20U 0204	4DA	M	WH	45,XY,-6/46,XY,r(6)	U	A	PROBAND			Multiple congenital anomalies; dysmorphic features: hydrocephaly, anal atresia, and congenital heart defect; 58% of cells show r(6) at passage 5
CHROMOSOME 07													
*GM3913	12	EEMEM	20I 0356	20MO	M	WH	46,XY/47,XY,+7	U	A	PROBAND			French Canadian; absent radii and short ulnae; microphthalmia; corneal opacity; chromatid breaks and gaps; centromere splitting and puffing; Roberts syndrome-SC Phocomelia; 46% 46,XY; 48% tetraploid at P15
CHROMOSOME 08													
GM0425	12	R1640	20U 0013	10FW	M		47,XY,+8,inv(9)(pter>p11::q21>p11::q21>qter)	U	A	PROBAND			Spontaneous abortion
*GM0496	3	EEMEM	20U 0056	2	M		46,XY/47,XY,+8	U	A	PROBAND			Mosaic; in fibroblasts, 81% are 47,XY,+8; in lymphocytes, 83% are 47,XY,+8
*GM2030	7	EEMEM	20U 0030	23	M	WH	46,XY/47,XY,+8	U	A	PROBAND			In fibroblasts, 58% have +8; mental retardation
GM2596	6	EEMEM	20U 0073	10MO	F	BL	46,XX/47,XX,+8	U	A	PROBAND			58% trisomy 8; mental retardation and failure to thrive

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 08													
GM4610		EEMEM	20U 0080		F		46,XX,dic(14;21)(14qter>14p13::21p13>21qter),+8/47,XX,+8	U	A	PROBAND			Additional t(14;21) present in only 25% of cells at passage 6; passage 3 at IMR
CHROMOSOME 09													
*GM2329	7	EEMEM	10U 0039	18DA	F	WH	46,XX/47,XX,+9	U	A	PROBAND			10% of fibroblasts show the +9; small for age, small upslanting palpebral fissures, micrognathia, & clinodactyly
*GM3226	2	EEMEM	20U 0081	9MO	M	BL	47,XY,+del(9)(9pter>9q11:)mat	U	A	PROBAND	602	1	Multiple malformations & mental retardation; see GM5067 Lymphoid
GM3225	4	EEMEM	20U 0326	32	F	BL	46,XX/47,XX,+del(9)(9pter>9q11:)	U	A	MOTHER	602	2	36% 46,XX in lymphocytes, 98% 46,XX in fibroblasts; short stature; mentally slow; see GM5066 Lymphoid
CHROMOSOME 10													
*GM1429	8	EEMEM	20U 0068	55	F	BL	47,X,+10,t(X;9)(Xpter>Xq13::9q34>9qter;9pter>9q34::Xq13>Xqter)	U	A	MOTHER	140	2	Mosaic; 16% of fibroblasts do not have additional 10; in blood leukocytes, normal X was late replicating; normal phenotype; fibroblasts have 164% of normal HK1 activity and 122% of normal GOT1 activity
CHROMOSOME 13													
GM0503	2	EEMEM	20U 0066	1	M		46,XY/47,XY,+13	U	A	PROBAND			54% of cells are 46,XY
*GM0526	3	EEMEM	20U 0095	NB	M	WH	47,XY,+13	U	A	PROBAND			Died shortly after birth; slow growing culture
GM2948A	4	EEMEM	20U 0107	NB	M	WH	47,XY,+13	U	A	PROBAND			Microcephaly, cebocephaly, microphthalmia, & polydactyly
GM3084A	12	EEMEM	20U 0224	11MO	M	BL	47,XY,+13	U	A	PROBAND			Multiple congenital anomalies
GM3330	12	EEMEM	20U 0044	1DA	M	WH	47,XY,+13	U	A	PROBAND			Multiple congenital anomalies
CHROMOSOME 14													
GM3233	2	EEMEM	20U 0122	1WK	M	WH	45,XY,-14/46,XY,r(14)	U	A	PROBAND			Congenital malformations; 88% of cells are 46,XY,r(14) at passage 4

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REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 15													
GM0682		EEMEM	20U 0013	30DA	F		46,XX/47,XX,+15	U	A	PROBAND			64% of cells are 46,XX; all cells have one 9qh+; spontaneous abortus; passage 2 at IMR
GM3133	6	EEMEM	20U 0273	10FW	F		46,X,+15	U	A	PROBAND			Culture established from a spontaneous abortus; grows slowly; 141% of normal pyruvate kinase-3 activity
*GM3184	5	EEMEM	20U 0273	11FW	M		47,XY,+15	U	A	PROBAND			Spontaneous abortus; 162% of normal pyruvate kinase-3 activity
GM7189	5	EEMEM	20U 0015	12FW	A	WH	47,XY,+15	U	A	PROBAND			Fibroblast culture established from fetal membranes
CHROMOSOME 16													
*GM4435	4	EEMEM	20U 0312	19FW	A	IN	48,XY,+16,+21	U	A	PROBAND			Spontaneous abortus; amnion fibroblast culture
CHROMOSOME 18													
*GM0143	3	EEMEM	20U 0107	3MO	F	BL	47,XX,+18	U	A	PROBAND			Died at age 3 months
GM0734	3	EEMEM	20U 0095	18FW	F		47,XX,+18	U	A	PROBAND			
GM1118	8	EEMEM	20U 0150	30MO	F	WH	45,XX,-18/46,XX,r(18)	U	A	PROBAND			78% of cells have r(18) at passage 18; multiple congenital anomalies
*GM1359	3	EEMEM	20U 0107	4WK	M	WH	47,XY,+18	U	A	PROBAND			
GM2732	7	EEMEM	20U 0109	4DA	M	WH	47,XY,+18	U	A	PROBAND			Classical syndrome findings
GM3538	2	EEMEM	20U 0107	NB	F	WH	47,XX,+18	U	A	PROBAND			Classical syndrome findings
GM3623	6	EEMEM	20U 0340	NB	F	BL	48,XXX,+18	U	A	PROBAND			Clinical features of variant trisomy 18
CHROMOSOME 20													
GM7408		EEMEM	20U 0426	12FW	F	OT	47,XX,+20	U	A	PROBAND			Oriental/Caucasian; culture established from membrane and chorionic villi of a spontaneous abortus

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME 21													
*GM0230B	14	EEMEM	20U 0088	14MO	F	WH	45,XX,-21	U	A	PROBAND			60% of normal superoxide dismutase-1; responds less than normal diploid fibroblasts to B- adrenergic stimulation; 12% of cells show random chromosome loss
*GM1137	6	R1640	10U 0017	1MO	M	BL	48,XXY,+21,t(6;10)(6qter>6p25::10p11> 10pter;10qter>10p11::6p25>6pter)	U	A	PROBAND			Down's syndrome features
*GM2067A	7	EEMEM	20U 0095	20FW	M	WH	47,XY,+21	U	A	PROBAND			Down's syndrome phenotype; same sensitivity to interferon A & B as normal diploid fibroblasts using both VSV and EMC virus; hyper-responsive to B- adrenergic stimulation
*GM2571	4	EEMEM	20U 0148	3MO	F	WH	48,XX,+21,+mar	U	A	PROBAND			Down's syndrome phenotype; 10 times more sensitive to A & B interferon (VSV virus) than normal diploid fibroblasts; equal sensitivity with EMC virus
*GM2767B	7	EEMEM	20U 0107	14	F		47,XX,+21	U	A	PROBAND			Down's syndrome phenotype
*GM3576	10	EEMEM	20U 0326	13FW	M	WH	48,XY,+2,+21	U	A	PROBAND			Spontaneous abortus
*GM4435	4	EEMEM	20U 0312	19FW	A	IN	48,XY,+16,+21	U	A	PROBAND			Spontaneous abortus; amnion fibroblast culture
GM4592A	3	EEMEM	20U 0107		F	WH	47,XX,+21	U	A	PROBAND			Down's syndrome phenotype
GM4614	6	EEMEM	20U 0260	2DA	F	WH	47,XX,+21	U	A	PROBAND			Down's syndrome phenotype
GM4616	5	EEMEM	20U 0260	3DA	F	WH	47,XX,+21	U	A	PROBAND			Down's syndrome phenotype
GM4617	5	EEMEM	20U 0260	1DA	F	WH	47,XX,+21	U	A	PROBAND			Puerto Rican; Down's syndrome phenotype
GM4965	7	EEMEM	20U 0271	1MO	M	OR	48,XXY,+21	U	A	PROBAND			Down's syndrome phenotype
*GM0260	6	EEMEM	20U 0095	11	M	WH	46,XY/47,XY,+21/47,XY,+t(21;21) (qter>cen>qter)	U	A	PROBAND	327	1	66%/24%/10% Mosaic; formerly GM0069

CHROMOSOMALLY ABERRANT HUMAN FIBROBLASTS

REF#	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM#	REMARKS
ANEUPLOID													
CHROMOSOME 21													
*GM2504E	9	EEMEM 20U	0095	1MO	M	BL	47,XY,+21	U	A	PROBAND	329	1	Formerly GM0258; shows increased sensitivity to interferon; Down's syndrome phenotype; hyper-responsive to B- adrenergic stimulation
*GM0137A	10	EEMEM 20U	0075	6	M	WH	45,XY,-21	U	A	PROBAND	335	1	60% of normal superoxide dismutase-1; in some cells, chromosome #4 shows extra unidentifiable material which may be from chromosome #21; multiple malforrations & severe psychomotor retardation
*GM0201	10	EEMEM 20U	0030	13	F	WH	47,XX,+21,t(1;17)(1qter>1p32::17p13> 17pter;17qter>17p13::1p32>1pter)	U	A	PROBAND	337	1	Down's syndrome phenotype
*GM1920	2	EEMEM 20U	0061	23	M	WH	47,XY,+21,inv(9)(pter>p11::q13>p11:: q13>qter)mat,t(8;14)(8pter>8q11::14q11> 14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	Down's syndrome phenotype; see GM1921A Lymphoid
GM4928	6	EEMEM 20U	0061	27	M	WH	47,XY,+21	U	A	PROBAND	781	1	Down's syndrome phenotype; 47,XY,+21, fra(16)(q22) in leukocytes; no fra(16) was observed in fibroblasts; see GM4927B Lymphoid
GM2524	7	EEMEM 20U	0148	10WK	M	WH	47,XY,+21/46,XY,-21,+dic(21)(pter> q21::p12>qter)	U	A	PROBAND	822	1	82% of cells are 47,XY,+21 at passage 10; small head, high arched palate, flat occiput, upslanting palpebral fissures, bilateral epicanthal folds, hypotonic fingers, & developmental delay
CHROMOSOME 22													
GM7106	5	EEMEM 20I	0426	8FW	M	OT	47,XY,+22s+	U	A	PROBAND			Hawaiian-Caucasian-Japanese; culture established from membrane and chorionic villi of a spontaneous abortus
TERATOMA CELL LINES													
CHROMOSOME 00													
*GM1304		EEMEM 20U	0056	27	F		46,XX	B	A	PROBAND	154	1	Passage 4 at IMR; PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/+;13=+/+; 15=+/+;21=+/+; right ovarian teratoma; same pt. as GM1305

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TERATOMA CELL LINES						
CHROMOSOME 00													
*GM1305		EEMEM	20U	0056	27	F	46,XX	B	A	PROBAND	154	1	Passage 4 at IMR; PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/-;13=+/-; 15=+/-;21=+/-; uterine tissue; 8% of cells show random chromosome loss; same pt. as GM1304
*GM1306A		EEMEM	20U	0056	31	F	WH 46,XX	B	A	PROBAND	155	1	Passage 8 at IMR; PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms: 13=-/-;14=+/+; 21=-/-; right ovarian teratoma; same pt. as GM1307
*GM1307		EEMEM	20U	0056	31	F	WH 46,XX	B	A	PROBAND	155	1	Passage 5 at IMR; PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms: 13=+/-;14=+/-; 21=+/-; fallopian tube tissue; 10% of cells show random chromosome loss; same pt. as GM1306A

Virus-Transformed Fibroblast Cultures

Virus-Transformed Fibroblast Cultures

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMMB	REMARKS
Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890												
GM5849	90	EEMEM 10U	0135	18	M		--		PROBAND	868	1	AT5BIVA; SV40 transformed GM5823; polyploid; radiosensitive
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
*GM2894	47	EEMEM 10U	0114	9	F		--	B	PROBAND			SV40 transformed culture, aneuploid; accumulates large amounts of intracellular nonprotein cystine
Fanconi Anemia (Pancytopenia) - 22765, 22766												
GM6914A	53	EEMEM 10U	0135	12	M	BL	--		PROBAND	969	1	SV40 transformed GM1309; clinically affected; lymphocytes show multiple chromosome breaks; untrans. fibro. show high freq. of spont. chromo. aberr. & extreme sensitivity to cell killing by Mitomycin-C; T-antigen positive
GM6935	21	EEMEM 10U	0135	12	F	WH	--		PROBAND	973	1	Arab; clinically affected; SV40 transformed culture; see GM2053 Fibroblast; untrans. fibro. are only slightly sensitive to cell killing by Mitomycin-C; modal no. of chromosomes = 114, range is 106 to 122; struct. aberr. & polyploidy
Galactosemia (Transferase Deficiency) - 23040												
*GM0639	46	EEMEM 20U	0089	8	M	BL	--	A	PROBAND	26	1	SV40 transformed GM0052; Gal-1-P uridyl transf. absent after crisis, T-antigen positive; polyploid; see also GM1908 Fibroblast & GM2796 Lymphoid
*GM0638	48	EEMEM 20U	0089	3MO	M	WH	--	A	PROBAND	27	1	SV40 transformed GM0054; Gal-1-P uridyl transf. absent post-crisis, G6PD type B; T-antigen positive; hypodiploid; see GM1907 Fibroblast & GM2795 Lymphoid
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
*GM0847		EEMEM 20U	0085	5	M	BL	Y-	A	AFF MAT COU	71	6	Line LN-SV; SV40 transformed GM2063; G6PD type A; > 95% of cells are T-antigen positive; HPRT deficient; two affected brothers; passage 9 at IMR
Apparently Normal: Non-Fetal Tissue												
*GM0637B	2	EEMEM 15U	0089	18	F	WH	++		PROBAND	162	2	SV40 transformed GM0037A (Puerto Rican); T-antigen positive; 43% of cells are diploid, 57% are tetraploid; all cells have t(7;7)(p22;q22) and random abnormalities
Osteogenic Sarcoma - 25950												
*GM5887		EEMEM 20U	0028	13	F	WH			PROBAND			Line 143B; human osteosarcoma cells transformed by murine sarcoma virus; deficient in thymidine kinase; tumorigenic in nude mice; passage 1 at IMR; modal no. of 50 chromosomes; all cells XO; 64% are polyploid

VIRUS-TRANSFORMED FIBROBLAST CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
Retinoblastoma: Sporadic												
GM3022	49	EEMEM 20U	0135	27MO	F	WH	+?	A	PROBAND	187	1	Bilateral; SV40 transformed GM1142; 100% T-antigen positive, 1-5% V-antigen positive; post-crisis line near triploid; multiple rearrangements; all cells have deleted chromosome 13; see GM1484 Lymphoid and GM1142A Fibroblast
Xeroderma Pigmentosum: Complementation Group A - 27870												
*GM4429B		EEMEM 10U	0111	7	F	WH	--		PROBAND	484	1	XP12BE; SV40 transformed; passage 5 at IMR; see GM2250A Lymphoid and GM5509 Fibroblast; less than 2% of normal UV induced unscheduled DNA synthesis in fibroblasts; T-antigen positive
*GM4312A		EEMEM 10U	0335	7	F	OR	--	B	PROBAND	695	1	Japanese; XP20S; SV40 transformed; severe XP; mental retardation; gait disturbance; see GM2345A Lymphoid; increased chromosome breakage; < 2% of normal UV induced unscheduled DNA synthesis; passage 3 at IMR; T-antigen positive

Apparently Normal Human Lymphoblasts

Apparently
Normal
Human
Lymphoblasts

APPARENTLY NORMAL HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
*GM3299	25HR	R1640 20I	0309	8	F		PROBAND			46,XX; negative for T cell 65,000 M.W. glycoprotein; positive for sIg and Ia (B cell) antigen; 6% of cells show random chromosome loss
GM3714	17HR	R1640 15I	0343	10	F	WH	PROBAND	391	1	Sister of GM3715; 46,XX; 1 cell out of 50 was 47,XX,+7
GM3798	21HR	R1640 20I	0343	10	M	WH	BRO	550	2	46,XY; 12% of cells show random chromosome loss; brother of GM3797
*GM0892A	35HR	R1640 20I	0024	12	F	WH	PROBAND			HLA type A2(A9),B12,B18; 45,XX,-18, der(12),t(12;18)(12qter>12p11::18q11> 18qter)/47,XX,+12; 78%/22%
GM3715	24HR	R1640 15I	0343	12	F	WH	SIS	391	2	46,XX; 10% of cells show random chromosome loss; sister of GM3714
GM3797	23HR	R1640 20I	0343	14	F	WH	PROBAND	550	1	46,XX; 12% of cells show random chromosome loss/gain; sister of GM3798
GM3469	23HR	R1640 20I	0120	20	M	WH	FATHER	239	1	46,XY; Mexican-American; daughter is GM3470 Lymphoid; 8% of cells show random chromosome loss
GM0946B	35HR	R1640 15I	0024	22	F	WH	PROBAND			46,XX; 18% of cells show random loss of 1 chromosome and 10% of cells show random loss of 2 chromosomes
*GM1953A	23HR	R1640 15I	0089	22	F	WH	PROBAND	360	1	Formerly GM1079; 46,XX; 10% of cells show random chromosome loss; 2% show random gain
GM2254	34HR	R1640 20I	0155	22	F	WH	PROBAND			46,XX/46,XX,9p+; 58%/42% in lymphoid culture; 46,XX in leukocytes
*GM0131	26HR	R1640 20I	0010	23	F	WH	PROBAND			46,XX; 4% of cells show random loss of 1 chromosome
*GM0333B	34HR	R1640 15I	0010	23	F	WH	PROBAND			46,XX; 8% of cells show random chromosome loss
*GM0546	38HR	R1640 20I	0089	23	F	WH	PROBAND			Spontaneously transformed; 46,XX; 10% of cells show random chromosome loss/gain
*GM0607A	26HR	R1640 20I	0025	23	F	WH	PROBAND			46,XX,19p+; secretes IgM(Kappa)
*GM0922A	43HR	R1640 15I	0024	23	F	WH	PROBAND	596	1	46,XX with 20% of cells showing random chromosome loss/gain; same patient as GM0924

APPARENTLY NORMAL HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
*GM0130B	20HR	R1640 15I	0010	25	M	WH	PROBAND	700	1	46,XY; negative for T cell 65,000 M.W. glycoprotein; positive for sIg and Ia (B cell) antigen; 8% of cells show random chromosome loss
*GM0558	27HR	R1640 20I	0089	26	M	WH	PROBAND			HLA type A3,A11,Bw15,Bw22; 46,XY; 6% of cells show random chromosome loss
GM1805	23HR	R1640 20I	0089	26	F	WH	PROBAND	355	1	HLA type A1,A9,B18(B13); formerly GM1073; 46,XX; 12% of cells show random chromosome loss
*GM3201	25HR	R1640 20I	0208	26	M	WH	PROBAND			Clinically normal; absent neutral alpha-glucosidase C activity; 46,XY; 10% of cells show random chromosome loss
*GM0536	40HR	R1640 20I	0089	27	M	WH	PROBAND			46,XY; 10% of cells show random chromosome loss
*GM0621A	26HR	R1640 15I	0089	28	M	WH	PROBAND			46,XY; 14% of cells show random chromosome loss/gain
*GM1078A	30HR	R1640 20I	0089	30	F	WH	PROBAND			46,XX; 20% of cells show random chromosome loss; 6% show random gain; centromeres of chromosomes #1 and #16 show fragility
GM0893A	26HR	R1640 15I	0024	32	F	WH	PROBAND	595	1	Same patient as GM0894; 46,XX with 24% of cells showing random chromosome loss (45) and 4% with random chromosome gain (47)
GM1806	32HR	R1640 20I	0089	33	M	WH	PROBAND	356	1	Formerly GM1074; HLA type A1,A2,Bw17,B18,(Bw22); 46,XY with 14% of cells showing random loss of 1 chromosome and 6% showing random loss of 2 chromosomes
*GM1989	19HR	R1640 20U	0089	33	M	WH	PROBAND	358	1	46,XY; 8% of cells show random chromosome loss; formerly GM1076
*GM2131A		R1640 15I	0205	33	M	WH	PROBAND			ATCC CCL 156; 45,X,1p+/46,XY; (62%/38%); karyotype is unstable with random chromosome loss and gain; G6PD type B; HLA type A2,w33;B7,14; secretes IgM(Lambda chains); line RPMI 1788
*GM2184A	27HR	R1640 15I	0186	36	M	WH	PROBAND	455	1	Normal spouse of HD patient; 46,XY/45,XY,-22; 50%/50%; 1 cell out of 50 showed 47,XY,+18; see GM2185 Fibroblast
GM6315		R1640 15I	0148	37	M	WH	PARENT	951	1	Father of a child with an unbalanced translocation; 46,XY; 4% of cells show random chromosome loss; wife (46,XX, t(11;16)) is GM6316A Lymphoid

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	REL TO PROBAND	FAM#	FAMM#	REMARKS
GM1815	52HR	R1640 20I	0089	42	M	WH	PROBAND	357	1	Formerly GM1075; HLA type A2,A9,B7,B13,(B12); 46,XY; 12% of cells show random chromosome loss
GM1814A	45HR	R1640 15I	0089	44	F	WH	PROBAND	354	1	Formerly GM1072; same pt. as GM1814 only seven years older; 46,XX; 20% of cells show random chromosome loss
GM1954	24HR	R1640 20I	0089	44	F	WH	PROBAND	361	1	Formerly GM1080; 46,XX; 24% of cells show random chromosome loss; 2% show random gain
GM5920	39HR	R1640 15I	0026	48	F	WH	PROBAND	887	1	46,XX; 14% of cells show random chromosome loss/gain; see GM5879 Fibroblast
*GM1310B		R1640 15I	0089	51	M	WH	PROBAND	476	1	See GM0967A Fibroblast; 46,XY; 8% of cells are tetraploid; 26% show random chromosome loss
GM1990	39HR	R1640 20I	0089	51	F	WH	PROBAND	359	1	46,XX/47,XX,+12; 36%/64%; formerly GM1077
GM1310A	32HR	R1640 15I	0089	57	M	WH	PROBAND	476	1	46,XY; 10% of cells show random chromosome loss; same as GM1310B only 6 years older; see GM0967B Fibroblast
*GM1056A	20HR	R1640 20I	0089	65	M	WH	PROBAND			HLA type A9,29,B12,Bw15; secretes IgA2 (Lambda); 46,XY; 36% of cells show random chromosome loss
GM3657	15HR	R1640 20I	0026	68	M	WH	PROBAND	542	1	46,XY; 10% of cells show random chromosome loss; see GM3658 Fibroblast

Biochemically Mutant Human Lymphoblasts

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Argininemia (Arginase Deficiency) - 20780												
*GM2011	28HR	R1640 20I	0006	5	F	WH	--		PROBAND	422	1	Mexican-American; see also GM0954 Fibroblast; RBC Arginase deficiency
Argininosuccinicaciduria (Argininosuccinate Lyase Deficiency) - 20790												
GM3010	21HR	R1640 20I	0207	8	F		--		PROBAND			Absent RBC Argininosuccinic acid lyase
Carnosinemia - 21270												
*GM2631	17HR	R1640 20I	0008	12	M	WH	--		PROBAND	363	1	46,XY; 14% of norm. plasma Carnosinase activity; profound mental retardation; severe athetoid spastic quadriparesis; optic atrophy; sensory peripheral neuropathy
Citrullinuria (Citrullinemia) - 21570												
GM1204	34HR	R1640 20I	0107	8MO	M	WH	--		PROBAND	2	1	HLA type A2,B12/A28,Bw35; fibroblasts are deficient in Argininosuccinate synthetase activity & show no detectable enzyme antigen; see GM1044 Fibroblast
*GM1206	35HR	R1640 20I	0107	27	M	WH	+-(0)		FATHER	2	2	See GM1058 Fibroblast; HLA type A11,Bw15/A2,B12; Neutral Alpha-glucosidase C phenotype=1; fibroblasts have 38% of normal Argininosuccinate synthetase activity
*GM1205	20HR	R1640 20I	0107	26	F	WH	+-(0)		MOTHER	2	3	HLA type A28,Bw35/A1,B13; Neutral Alpha-glucosidase C phenotype=1
*GM1207	28HR	R1640 20I	0107	5	F	WH	+?		SIS	2	4	HLA type A11,Bw15/A28,Bw35; possible heterozygote
GM1685	24HR	R1640 20I	0157	1DA	M	WH	--	A	PROBAND	3	1	See GM1679 Fibroblast; HLA type A1,Aw32,B18,B27; deficient Argininosuccinate synthetase; a sib died of citrullinuria
*GM0235	96HR	R1640 20I	0010	31	M	WH	--	B	PROBAND	699	1	HLA type (Aw31,33)A9,Bw15,B12; deficient Argininosuccinate synthetase; 46,XY; line UM-21
Cystathioninuria - 21950												
GM1454	88HR	R1640 20I	0008	7	M	OR	--		PROBAND			Unresponsive to B6; HLA type A28,A2,Bw40,Bw21; 92% of cells are 47,XY,+12; 68% of cells show 13q+; random chromosome rearrangements are present
GM1562	18HR	R1640 20I	0008	42	M	WH	+-(0)		PARENT			HLA type A3,A10,B13,B18; proband not in Repository; 46,XY; 22% of cells show random chromosome loss

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM											
Cystathioninuria - 21950											
GM1781	23HR	R1640 20I	0008	18	F	WH	--	PROBAND	169	1	B6 responsive; 46,XX; 4% of cells show random chromosome loss
GM1461	38HR	R1640 20I	0008	39	F	WH	+-(O)	MOTHER	169	2	HLA type A2,A3,Bw22,Bw40; 46,XX; 40% of cells show random chromosome loss/gain
GM1565	35HR	R1640 20I	0008	16	F	WH	--	PROBAND	170	1	Also has PKU; HLA type A28,Aw24,B7,Bw35
GM1807	40HR	R1640 20I	0008	--	M	WH	+-(O)	FATHER	170	2	HLA type A1,A2,Bw35,B13; clinically unaffected
GM1566	36HR	R1640 20I	0008	16	F	WH	--	PROBAND	406	1	HLA type A1,A28,B13,Bw35; 47,XX,+12/46,XX,-10,+12/45,XX,-10; 66%/30%/4%
GM1456A		R1640 15I	0008	43	F	WH	+-(O)	MOTHER	406	2	HLA type A1,B13,Bw27; 46,XX; 14% of cells show random chromosome loss/gain; clinically unaffected
GM1868	37HR	R1640 20I	0008	49	M	WH	+-(O)	FATHER	406	3	HLA type A28,A10,Bw15,Bw16; 46,XY; 20% of cells are polyploid; 10% of cells show random chromosome loss; clinically unaffected
Gyrate Atrophy of Choroid and Retina (Ornithinemia) - 25887											
*GM3534	44HR	R1640 20I	0334	37	F	WH	--	PROBAND			Portuguese; deficient Ornithine aminotransferase activity in PHA stimulated lymphocytes and fibroblasts
GM3540	21HR	R1640 20I	0334	36	F	WH	--	PROBAND			Scotch-Irish-English; deficient Ornithine aminotransferase activity in fibroblasts
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620											
GM1446	62HR	R1640 20I	0008	--	F	WH	+-(O)	PARENT			HLA type A2,3,Bw35,Bw17; proband not in Repository; clinically unaffected
GM1447	38HR	R1640 20I	0008	--	F	WH	+-(O)	PARENT			HLA type A1,A10,B7,(Bw16); proband not in Repository; clinically unaffected
GM1463	24HR	R1640 20I	0008	42	F	WH	+-(O)	PARENT			HLA type A2,A11,B27,Bw22; proband not in Repository; clinically unaffected
GM1528	18HR	R1640 20I	0008	--	M	--	+-(O)	PARENT			HLA type A10,(A3),B18,Bw22; proband not in Repository; clinically unaffected
GM1529	19HR	R16-0 20I	0008	43	F	--	+-(O)	PARENT			HLA type A10,Aw23,B8,B18; proband not in Repository; clinically unaffected

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF AMINO ACID METABOLISM												
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
GM1532	24HR	R1640 20I	0008		M		--		PROBAND			HLA type A2,B7,Bw22,(B18); clinically affected
GM1558	36HR	R1640 20I	0008		F		+-(O)		PARENT			HLA type A10,Aw24,B7,Bw15; proband not in Repository; clinically unaffected
GM1559	23HR	R1640 20I	0008		M		+-(O)		PARENT			HLA type A3,A10,Bw15,B27; proband not in Repository; clinically unaffected
GM1560	22HR	R1640 20I	0008		M		--		PROBAND			HLA type A2,3,B7,B18; clinically affected
GM1808	25HR	R1640 20I	0008	45	M	WH	+-(O)		PARENT			HLA type A2,A10,B12,B27; proband not in Repository; clinically unaffected
GM2689	20HR	R1640 20I	0038	25	M	WH	--		PROBAND	241	1	Dislocation of left optic lens; glaucoma of right eye; left sided seizures
GM2688A		R1640 15I	0038	27	M	WH	+?		BRO	241	2	Clinically normal; possible heterozygote
GM2690	28HR	R1640 20I	0038		M	WH	+-(O)		FATHER	241	3	Clinically unaffected
GM2429	25HR	R1640 20I	0008		F	WH	--		PROBAND	480	1	Formerly GM2375; clinically affected
Maple Syrup Urine Disease (Branched-Chain Ketoaciduria) - 24860												
GM1366	34HR	R1640 20I	0107	7	F	BL	--	A	PROBAND	20	1	See GM1364 Fibroblast; HLA type A2(A9),B8,B12; classical symptoms; one previously affected male sib; deficient branched chain keto-acid decarboxylase activity
GM1655	34HR	R1640 20I	0107	5	F	WH	--	A	PROBAND	21	1	See GM1654 Fibroblast; HLA type A1,A2,B13,Bw17(B23)(Bw40); Mennonite kindred; two female sibs died of MSUD; fibroblasts show branched chain keto-acid decarboxylase deficiency
Phenylketonuria (Phenylalanine Hydroxylase Deficiency) - 26160												
GM2659A		R1640 20I	0038	14	F	WH	--		PROBAND			Puerto Rican; profound mental retardation; spastic quadriparesis
DISORDERS OF CARBOHYDRATE METABOLISM												
Amyloidosis I (Andrade Type; Portuguese Type of Amyloid Neuropathy) - 10480												
*GM1999	27HR	R1640 20I	0155	16	F	WH	+-		PROBAND	421	1	Non-Portuguese; see GM1998 Fibroblast; 4 effected generations (all female); vitreous opacities & perivascular exudates; affected mother died at age 21

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Disaccharide Intolerance I (Congenital Sucrose-Isomaltose Malabs) - 22290												
GM3070	16HR	R1640 20I	0297	14	M	WH	--		PROBAND			Jejunal mucosa revealed no sucrase activity
Fucosidosis - 23000												
GM1025	24HR	R1640 20I	0008	4	M	WH	--	B	PROBAND	24	1	See GM0292 Fibroblast; deficient Alpha-L-Fucosidase; Type 2; similarly affected sib
GM1024	28HR	R1640 20I	0008	9	M	WH	--	A	AFF BRO	24	2	Type 2; see GM0291 Fibroblast; deficient Alpha-L-Fucosidase
*GM1026	22HR	R1640 20I	0008	29	F	WH	+-(O)	B	MOTHER	24	3	See GM0290 Fibroblast; low-normal Alpha-L-Fucosidase activity; Neutral Alpha-glucosidase C phenotype=1; two affected children
*GM1023	30HR	R1640 20I	0008	31	M	WH	+-(O)	B	FATHER	24	4	See GM0289 Fibroblast; low-normal Alpha-L-Fucosidase activity; enzyme phenotype: Neutral Alpha-glucosidase C=1, ADA=1,1; two affected children
Galactosemia (Transferase Deficiency) - 23040												
GM2796	26HR	R1640 20I	0260	15	M	BL	--		PROBAND	26	1	See GM0052, GM1908 Fibroblast & GM0639 (SV40 Transformed); no detectable transferase activity in RBCs or fibroblasts
GM2795	19HR	R1640 20I	0095	7	M	WH	--		PROBAND	27	1	See GM0054, GM1907 Fibroblast and GM0638 (SV40 Transformed); normal kinase and deficient transferase activity in fibroblasts
GM2412B		R1640 15I	0008	15	M	WH	--	B	PROBAND	28	1	Formerly GM1027; HLA type A2,B12/A1,B8; deficient galactose-1-phosphate uridyl transferase; similarly affected sib; see GM0441A Fibroblast; 46,XY in fibroblasts
GM1028	42HR	R1640 20I	0008	16	F	WH	--		AFF SIS	28	2	HLA type A1/A2,B12; see GM0440 Fibroblast; 46,XX/46,X,t(X;11); 92%/8%; balanced in fibroblasts; no detectable transferase activity in fibroblasts
GM1029	76HR	R1640 20I	0008	41	F	WH	+-(O)		MOTHER	28	3	HLA type A2,B12/A28,Bw35; see GM0439 Fibroblast; 50% of normal transferase activity in erythrocytes; two affected children; 46,X,t(X;3)(Xpter>Xq26::3p12>3pter;3qter>3p12::Xq26>Xqter); balanced in fibroblasts

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Galactosemia (Transferase Deficiency) - 23040												
GM0148	106HR	R1640 20I	0010	8	M	WH	--	B	PROBAND	171	1	46,XY; deficient galactose-1-phosphate uridyl transferase
GM0149	38HR	R1640 20I	0010	47	M	WH	+-(O)	B	FATHER	171	2	46,XY; reduced galactose-1-phosphate uridyl transferase
Glycogen Storage Disease: Type II - Pompe Disease - 23230												
GM6314		R1640 15I	0208	40	F	WH	--	A	PROBAND			Adult onset; respiratory arrest; muscle weakness; alpha-1,4 glucosidase deficiency: 1% of normal activity in lymphoblasts; similarly affected sister
GM1464	24HR	R1640 20I	0008	46	F	WH	--	B	PROBAND	173	1	Acid maltase deficiency; adult form
GM1778	30HR	R1640 20I	0008	16	F	WH	+-(O)		DAU	173	2	Obligate heterozygote - affected mother
GM1568	26HR	R1640 20I	0008	71	M	WH	+-(O)	B	FATHER	173	3	Heterozygote enzyme level of acid maltase
Glycogen Storage Disease: Type VII - Muscle PFK Deficiency - 23280												
*GM3128	18HR	R1640 20I	0317	31	M	WH	--	A	PROBAND			Tarui's disease; muscle, fibroblasts and lymphoblasts deficient in phosphofructokinase M subunits
GM7754		R1640 15I	0317	75	M	WH	--		PROBAND	2000	1	Atypical; late onset myopathy; 10 year history of proximal limb weakness without cramps, myoglobinuria, or hyperuricemia; 1% of normal muscle PFK activity & 55% of normal RBC PFK activity; mutant, unstable M unit in RBC
Mucolipidosis: Type II - I-Cell Disease - 25250												
GM2687		R1640 20I	0038	26	M	WH	+-(O)		PARENT	469	2	Father of an affected child; proband not in Repository
GM2660	23HR	R1640 20I	0008	23	F		+-(O)		PARENT	469	3	Mother of an affected child; proband not in Repository
Mucolipidosis: Type IV (Ganglioside Sialidase Deficiency) - 25265												
*GM2533	30HR	R1640 20I	0077	2	F	WH	--	B	PROBAND	229	1	Ashkenazi; see GM2048 Fibroblast; affected sib is GM2529 Amniotic; photophobia; bilateral corneal opacities; abnormal lysosomal storage bodies
Mucopolysaccharidosis: Type IH - Hurler Syndrome - 25280												
GM1034A		R1640 15I	0008	2	F	WH	--		PROBAND	599	1	HLA type A2,Aw30,B12,Bw22; same patient as GM1867; clinically affected

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CARBOHYDRATE METABOLISM												
Mucopolysaccharidosis: Type IH/S - Hurler/Scheie - 25280												
GM1032	28HR	R1640 20I	0008	15	M	IN	--		PROBAND			HLA type A9,A10,B13; clinically affected; Alpha-L-Iduronidase deficiency confirmed in skin fibroblasts
Mucopolysaccharidosis: Type IIIA - Sanfilippo Syndrome, A - 25290												
GM1780	36HR	R1640 20I	0008	8	M	WH	--		PROBAND			Clinically affected
Mucopolysaccharidosis: Type VI - Maroteaux-Lamy Syndrome - 25320												
*GM1022	31HR	R1640 20I	0008	4	F	WH	--	B	PROBAND	51	1	Deficient arylsulfatase B; short, coarse facies, prominent forehead, corneal opacities, lumbar kyphosis; stiff joints, umbilical hernia, mild hepatosplenomegaly, and dysostosis multiplex; see GM0519 Fibroblast; same pt. as GM5358
*GM5358		R1640 15I	0032	12	F	WH	--		PROBAND	51	1	Congestive heart failure; dysostosis multiplex; corneal clouding; deficient arylsulfatase B in fibroblasts and peripheral blood leukocytes; same patient as GM1022
GM3721	70HR	R1640 20I	0016	25	F	BL	+-(O)		MOTHER	392	1	See GM3720 Fibroblast; clinically unaffected
GM3723	34HR	R1640 20I	0016	40MO	F	BL	--		PROBAND	392	2	5% of normal arylsulfatase B activity; see GM3722 Fibroblast
GM3725	20HR	R1640 20I	0016	41	M	BL	+-(O)		FATHER	392	3	See GM3724 Fibroblast; clinically unaffected
GM3727	100HR	R1640 20I	0016	9	F	BL	+?		SIS	392	4	Possible heterozygote; see GM3726 Fibroblast
Neuraminidase and Beta-Galactosidase Deficiency - 25654												
*GM4305		R1640 15I	0126	8	F	WH	--		PROBAND	693	1	Infantile onset; visceromegaly; dysostosis multiplex; normal stature and intelligence; deficient beta-galactosidase and neuraminidase activity in fibroblasts; see GM0806 Fibroblast
*GM6091		R1640 15I	0126	40	F	WH	+-(O)		MOTHER	693	2	Normal beta-galactosidase and neuraminidase activity; see GM4307 Fibroblast
*GM6092A		R1640 15I	0126	40	M	WH	+-(O)		FATHER	693	3	Normal beta-galactosidase and neuraminidase activity; see GM4306 Fibroblast
Phosphoglycerate Kinase Deficiency - 31180												
*GM4535	17HR	R1640 15I	0380	15	M	OR	Y-		PROBAND			Japanese; PGK Tokyo; 16% of normal PGK activity; nonspherocytic hemolytic anemia and mental retardation

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Abetalipoproteinemia - 20010												
*GM1453	38HR	R1640 20I	0008	18	F	WH	--		PROBAND			Clinically affected
*GM1810	25HR	R1640 20I	0008	23	M	WH	--		PROBAND	398	1	HLA type A1,A10,B7,B13; formerly GM1451; clinically affected
Fabry Disease (Diffuse Angiokeratoma, Hereditary Dystopic Lipidosis) - 30150												
GM4391	15HR	R1640 15I	0032	41	M	WH	Y-		PROBAND			Variant; mild clinical manifestations; residual alpha-galactosidase A activity in fibroblasts and WBC; no activity in plasma
Farber Lipogranulomatosis; Ceramidase Deficiency - 22800												
*GM5748		R1640 15I	0175	5MO	F	WH	--		PROBAND	862	1	Hepatosplenomegaly; nodules on hands and feet; pulmonary infiltrates; anemia; hoarse cry; adenopathy; elev. liver ceramide level; undetect. acid ceramidase activity in leukocytes & fibroblasts; similarly affected bro.; see GM5752 Fib.
GM2 Gangliosidosis: Tay-Sachs Disease, AB Variant - 27275												
GM3691	26HR	R1640 15I	0328	13	F	WH	--	A	PROBAND			Dysarthria, dementia, & ataxia; normal B-hexosaminidase A & B, B-galactosidase, arylsulfatase A, & glycoprotein neuraminidase activities
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
GM2366	60HR	R1640 20I	0008	28	M	WH	+-	B	PROBAND			Carrier; heterozygous levels of hexosaminidase A
GM3441	19HR	R1640 20I	0328	37	F	WH	--?	A	PROBAND			Variant; hexosaminidase A deficiency; dementia; ataxia; upper & lower motor neuron involvement; Ashkenazi
GM3461	19HR	R1640 20I	0032	24	M	WH	--?		PROBAND			Ashkenazi; variant; deficient hexosaminidase A; weakness and ataxia
GM3575	26HR	R1640 20I	0328	31	F	WH	--?	A	PROBAND			Ashkenazi; ataxia, dementia; peripheral neuropathy; deficient hexosaminidase A; variant
GM3585	20HR	R1640 20I	0032	13MO	F	WH	--	B	PROBAND			5% of normal hexosaminidase A activity
GM3586	20HR	R1640 20I	0032	36MO	F	WH	--	B	PROBAND			8% of normal hexosaminidase A activity
GM3770	18HR	R1640 20I	0328	15	M	WH	--?	A	PROBAND	494	1	Jewish; variant; hexosaminidase A deficiency in white cells and lymphoblasts; mild dysarthria; three similarly affected sibs

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
GM2 Gangliosidosis: Type I - Tay-Sachs Disease - 27277, 27280												
GM3771	16HR	R1640 20I	0328	13	M	WH	--?	A	AFF BRO	494	2	Jewish; severe tremor, moderate ataxia, mild dysarthria; hexosaminidase A deficiency in white cells and lymphoblasts
GM3772	17HR	R1640 20I	0328	11	M	WH	--?	A	AFF BRO	494	3	Jewish; mild ataxia and hand tremors; hexosaminidase A deficiency in white cells and lymphoblasts; twin of GM3773
GM3773	19HR	R1640 20I	0328	11	M	WH	--?	A	AFF BRO	494	4	Jewish; mild ataxia and hand tremors; hexosaminidase A deficiency in white cells and lymphoblasts; twin of GM3772
Gaucher Disease: Type III (Juvenile and Adult, Cerebral) - 23100												
*GM1019	52HR	R1640 20I	0008	35	M	WH	+-(0)	B	PARENT			Father of a Juvenile Gaucher; low-normal acid B-glucosidase; proband not in Repository
GM1769B		R1640 20I	0008	25	M	WH	--	B	PROBAND	249	1	Adult Gaucher; formerly GM1021; low acid B-glucosidase
*GM1020	25HR	R1640 20I	0008	49	F	WH	+-(0)	B	MOTHER	249	2	Low-normal acid B-glucosidase
GM1030	30HR	R1640 20I	0008	50	F	WH	+-(0)	B	PARENT	250	1	Mother of a Juvenile Gaucher; low-normal acid B-glucosidase; proband not in Repository
GM1031	50HR	R1640 20I	0008	53	M	WH	+-(0)	B	PARENT	250	2	Father of a Juvenile Gaucher; low-normal acid B-glucosidase; proband not in Repository
Hepatic Lipase Deficiency - Hyperlipoproteinemia												
*GM6945		R1640 15I	0359	48	M	WH			PROBAND	976	1	Increased LDL and IDL; skin xanthomata; coronary heart disease with diabetes mellitus; positive family history; uncertain inheritance; see GM6946 Fibroblast
Combined Hyperlipidemia - 14425												
*GM1455A		R1640 15I	0008	33	M		+-		PROBAND			HLA type A11(A3),Bw22,(Bw40); clinically affected
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
*GM1458A		R1640 15I	0008	30	F		--		PROBAND			Receptor negative by clinical diagnosis
GM1460	35HR	R1640 20I	0008		F		+-		PARENT			Mother of a clinically diagnosed receptor negative child; proband not in Repository

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Familial Hypercholesterolemia - 14389 & Hyperlipoproteinemia: Type II-14440												
GM1567	34HR	R1640 20I	0008	28	M	BL	--		PROBAND			Abnormal regulation of 3-hydroxy-3-methylglutaryl coenzyme A reductase and incorporation of oleate into cholesteryl esters
*GM1767	34HR	R1640 20I	0008	19	F		--	B	PROBAND	264	1	Formerly GM1452; receptor negative
GM1784	24HR	R1640 20I	0008	23	M		--		PROBAND	265	1	Formerly GM1462; clinically affected
GM2373A	58HR	R1640 15I	0008	42	M	WH	+-		PARENT	271	7	Proband not in Repository; elevated serum cholesterol; two similarly affected children
GM1459A		R1640 15I	0008	25	F	WH	--	A	PROBAND	387	1	See GM0486 Fibroblast; lymphoid line is verified receptor negative
*GM1448	40HR	R1640 20I	0008	18	F	WH	+-		PROBAND	506	1	Elevated serum cholesterol; affected father and sister
GM2110	23HR	R1640 20I	0008	49	M	WH	+-		AFF FATHER	506	2	Formerly GM1450; tendon xanthoma; premature myocardial infarctions; elevated cholesterol levels (above 400 mg%); died before age 50
GM1766	38HR	R1640 20I	0008	23	F	WH	+-		AFF SIS	506	3	Formerly GM1449; elevated serum cholesterol
Krabbe Disease (Globoid Cell Sclerosis) - 24520												
GM6805		R1640 15I	0458	2	F	WH	--		PROBAND	962	1	Seizures; cortically blind; developmental delay; poor feeding; onset at age 6 months; deficient WBC lactosylceramide B-galactosidase activity; see GM6806 Fibroblast
GM6807A		R1640 15I	0458		M	WH	+-(O)		FATHER	962	2	Clinically normal; heterozygote level of lactosylceramide B-galactosidase activity in WBC; see GM6810 Fibroblast
GM6809		R1640 15I	0458		F	WH	+-(O)		MOTHER	962	3	Clinically normal; heterozygote level of lactosylceramide B-galactosidase activity in WBC; see GM6808 Fibroblast
Medium-Chain Acyl CoA Dehydrogenase Deficiency												
GM7523		R1640 15I	0187	17	F	WH	--		PROBAND	1058	1	Recurrent coma; dicarboxylic aciduria; less than 10% of normal medium-chain Acyl CoA Dehydrogenase activity in leukocytes; non-ketotic hypoglycemia; see GM6125 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF LIPID METABOLISM												
Metachromatic Leukodystrophy: Infantile (Arylsulfatase A Deficiency) - 25010												
*GM1018A		R1640 15I	0008	25	F	WH	+-(0)	B	PARENT			Mother of Infantile MLD; reduced activity of arylsulfatase A; produces IgG4; proband not in Repository; Puerto Rican
Metachromatic Leukodystrophy: Juvenile - 25020												
GM1017	99HR	R1640 20I	0008	14	F	WH	--	B	PROBAND	622	I	HLA type A2,A29,B12,B16; same patient as GM1785; deficient arylsulfatase A
GM1016	44HR	R1640 20I	0008	42	M	WH	+-(0)	B	FATHER	622	2	Carrier level of arylsulfatase A
Multiple Sulfatase Deficiency Disease - 27220												
*GM4916		R1640 15I	0175	4	M	WH	--	B	PROBAND			Severe mental retardation; dysostosis multiplex; ichthyosis rash; arylsulfatase A in fibroblasts 23% of normal, but less than 5% in urine, leukocytes, and serum; elevated urinary sulfatides and MPS
*GM3246	30HR	R1640 20I	0105	9	M	WH	--		PROBAND	632	1	See GM3245 Fibroblast; microcephaly, delayed psychomotor devel.; hypotonia, weakness, blind, deaf, and myoclonus; deficient arylsulfatase A,B,& C in leukocytes; excess mucopolysaccharides in urine (mostly heparan sulfate)
Niemann-Pick Disease: Type C (Sphingomyelinase Deficiency) - 25720												
*GM3124	23HR	R1640 20I	0107	9	F	WH	--		PROBAND	451	1	See GM3123 Fibroblast; 38% of normal sphingomyelinase activity and normal B-galactosidase activity in fibroblasts
Refsum Syndrome; Phytanic Acid Oxidase Deficiency - 26650												
*GM3897		R1640 15I	0175	38	M	WH	--		PROBAND	574	1	Elevated phytanic acid in blood, fat, & other body fluids and cultured skin fibroblasts; retinitis pigmentosa; see GM3896 Fibroblast
Sea-Blue Histiocyte Disease - 26960												
*GM1913	20HR	R1640 20I	0348	24	F	WH	--		PROBAND	407	1	See GM1912 Fibroblast; HLA type A2,Aw31, B18,B13; muscle biopsy showed foamy histiocytes & sea-blue histiocytes; eye movement abnormalities
MUTATIONS OF THE HEMOGLOBIN LOCI												
Beta Globin Region Restriction Polymorphisms												
GM5048	18HR	R1640 15I	0399	22	M	WH			PROBAND			In blood: Hpa I x beta: 7.6/7.6 Kb; Bam HI x beta: 8.3/8.3 Kb; Hind III x gamma (G,A): 7.1, 2.7/7.8, 3.4 Kb; Hinc II x pseudo B1: 7.6/6.0 Kb

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
MUTATIONS OF THE HEMOGLOBIN LOCI												
Beta Globin Region Restriction Polymorphisms												
GM5049B		R1640 15I	0399	22	M	WH			PROBAND			In blood: Hpa I x beta: 7.6/7.6 Kb; Bam HI x beta: 8.3/22.0 Kb; Hind III x gamma (G+A): 7.1, 2.7/7.8, 3.4 Kb; Hinc II x pseudo B1: 7.6/6.0 Kb
GM5050		R1640 15I	0399	35	F	WH			PROBAND			In blood: Hpa I x beta: 7.6/7.6 Kb; Bam HI x beta: 22/22 Kb; Hind III x gamma (G+A): 7.1, 3.4/7.1, 3.4 Kb; Hinc II x pseudo B1: 3.0/3.0 Kb
GM5051A		R1640 15I	0399	25	M	WH			PROBAND			In blood: Hpa I x beta: 7.6/7.6 Kb; Bam HI x beta: 8.3/22 Kb; Hind III x gamma (G+A): 7.8, 3.4/7.1, 3.4 Kb
GM5052	28HR	R1640 15I	0399	28	F	BL			PROBAND			In blood: Hpa I x beta: 7.6/7.0 Kb; Bam HI x beta: 8.3/8.3 Kb
Human Erythroleukemic Cell Lines												
*GM6141A		R1640 12U	0439	30	M	WH		B	PROBAND			Line HEL; derived from the peripheral blood of a pt. with Hodgkin's disease who later developed erythroleukemia; neg for EBVNA, immunoglobulins & T-lymphoid cell surface antigens; produces mainly Hb Barts in response to hemin
*GM5372C		R1640 15I	0412	53	F	WH		B	PROBAND	722	1	Line K562; establ. from a patient with chronic myelogen. leukemia by Lozzio & Lozzio; produces embryonic hemoglobin in response to hemin; modal no. of chromo = 60; range is 55-62; struct. aberrat. for chromos #1,#3,#9,#13,& #14 were observed
Thalassemias - 27350 and Hereditary Persistence of Fetal Hemoglobin - 14247												
GM7405		R1640 15I	0399	14	M	WH			PROBAND			B-thalassemia: homozygous for IVS-1 #110
GM7425		R1640 15I	0399	12	M	WH		A	PROBAND			Mediterranean B-thalassemia; IVS-1 #110 B+; nonsense 39 B-zero
*GM7426		R1640 15I	0399	4	M	OR			PROBAND			Chinese; B-thalassemia; C to T substitution at IVS-2 #654; insertion of A between codons 71 and 72
*GM7428		R1640 15I	0399	33	M	WH			PROBAND			Greek; B-thalassemia; IVS-2 #745 B+ (detectable with Rsa I); IVS-1 #6 (donor consensus sequence)
*GM2064	20HR	R1640 20I	0192	53	M	BL	--	A	PROBAND			Ghanaian; homozygous for hereditary persistence of fetal Hb, heterozygous for elliptocytosis; both G-gamma & A-gamma chains are synthesized; delta & beta globin gene DNA are deleted; 46,XY

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
MUTATIONS OF THE HEMOGLOBIN LOCI												
Thalassemias - 27350 and Hereditary Persistence of Fetal Hemoglobin - 14247												
GM7444		R1640 15I	0399	15	F	WH		A	PROBAND			Greek; B-thalassemia; homozygous for IVS 2 #1 B-zero (detectable with endonuclease Hph I)
GM7445		R1640 15I	0399	16	M	WH		A	PROBAND			Italian-Irish; B-thalassemia; IVS-2 #1 (detectable with endonuclease Hph I) B-zero, nonsense 39
*GM7905		R1640 15I	0523	34	F	BL			PROBAND			Heterozygous for B-thalassemia minor and hemoglobin J (Altgeld Gardens); patient is only slightly anemic
*GM2232	24HR	R1640 20I	0192	48	F	WH	+-		PROBAND	470	1	Greek; double heterozygote for hereditary persistence of fetal hemoglobin and B-thalassemia trait; 46,XX; 24% of cells show random chromosome loss; 15% are polyploid; A-gamma globin chain synthesis predominates
*GM2266A		R1640 15I	0192	17	F	WH	--	A	PROBAND	514	1	Sicilian; homozygous delta-beta thalassemia; similarly affected sib
*GM2267	26HR	R1640 20I	0192	19	F	WH	--	A	AFF SIS	514	2	Sicilian; homozygous delta-beta thalassemia; the 3' breakpoint is located approximately 5 kb to the 3' side of the B-globin gene; the overall deletion comprises approximately 13 kb
*GM2568	27HR	R1640 20I	0192		M		--	A	PROBAND	520	1	Homozygous for hemoglobin Lepore; 46,XY; 4% of cells show random chromosome loss or gain
*GM7406		R1640 15I	0399	28	F	WH			PROBAND	1039	1	Mediterranean B-thalassemia: -87 / IVS-1 #110 (B+)
*GM7407A		R1640 15I	0399	33	M	WH			AFF SPOUSE	1039	2	Mediterranean B-thalassemia: IVS-1 #6 / IVS-1 #1; Italian
DISORDERS OF METAL METABOLISM												
Menkes Syndrome (Kinky Hair Disease) - 30940												
GM1245	26HR	R1640 20I	0107	5DA	M	WH	Y-		AFF BRO	79	2	See GM1057 Fibroblast; HLA type A9,A10, B13,B18; fibroblasts show increased copper concentration; proband is GM0220 Fibroblast
GM1982	20HR	R1640 20I	0107	2	M	WH	Y-		PROBAND	80	1	See GM1981 Fibroblast; fibroblasts show elevated copper concentration

REP #	GEN \TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF METAL METABOLISM												
Menkes Syndrome (Kinky Hair Disease) - 30940												
GM1984	19HR	R1640 20I	0107	26	F	WH	+?		MOTHER	80	2	See GM1983 Fibroblast; son is only affected child in an extended family
Wilson Disease (Hepatolenticular Degeneration) - 27790												
GM5258		R1640 15I	0409	34	F	WH	--		PROBAND	841	1	Diagnosed at age 24 as asymptomatic with "emotional instability"; low serum ceruloplasmin & high hepatic copper level; maintained on d-penicillamine; father had Wilson's disease; parents are first cousins; see GM5257 Fibroblast
GM5256		R1640 15I	0409	59	F	WH	+-(O)		MOTHER	841	2	Clinically unaffected; two affected children; see GM5255 Fibroblast
GM5260		R1640 15I	0409	30	F	WH	--		AFF SIS	841	3	Diagnosed at age 19 with "hepatitis"; maintained on d-penicillamine with resolution of liver disease & no evidence of CNS dysfunction; see GM5259 Fibroblast
GM5340		R1640 15I	0409	25	F	WH	--		PROBAND	848	1	Onset at age 13 with jaundice, evidence of chronic liver disease, presence of Kayser-Fleischer rings, & low ceruloplasmin; controlled with d-penicillamine with no evidence of liver or CNS dysfunction; see GM5339 Fibroblast
GM5342		R1640 15I	0409	23	F	WH	--		AFF SIS	848	2	Diagnosed as asymptomatic at age 12 with low serum ceruloplasmin, elevated urinary & hepatic copper levels; maintained on d-penicillamine with no evidence of liver disease or CNS dysfunction; see GM5341 Fibroblast
GM5761A		R1640 15I	0409	53	F	WH	+-(O)		MOTHER	848	3	No evidence of Wilson's disease; rheumatoid arthritis since age 21; two affected daughters; see GM5762 Fibroblast
GM5367		R1640 15I	0409	32	F	WH	--		PROBAND	850	1	First diagnosed at age 25; low ceruloplasmin, hypersplenism, post-necrotic cirrhosis with portal hypertension and fluid retention, and elevated urinary copper excretion; on penicillamine; see GM5366A Fibroblast
GM5371		R1640 15I	0409	70	M	WH	+-(O)		FATHER	850	3	Clinically unaffected; see GM5370 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF METAL METABOLISM												
Wilson Disease (Hepatolenticular Degeneration) - 27790												
GM5797		R1640 15I	0409	37	F	WH	--		PROBAND	865	1	Diagnosed at age 25; neurological abnormalities; Kayser-Fleisher rings; low serum ceruloplasmin; inc. urinary copper excretion; elev. hepatic copper level; no clinical features of liver disease; on penicillamine; see GM5798 Fibroblast
GM5793		R1640 15I	0409	75	M	WH	+-(0)		FATHER	865	2	History of arthritis and coronary artery disease; see GM5794 Fibroblast
GM5795		R1640 15I	0409	70	F	WH	+-(0)		MOTHER	865	3	Clinically unaffected; see GM5796 Fibroblast
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenine Phosphoribosyltransferase Deficiency - 10260												
GM7290		R1640 15I	0497	26	F	WH	--		PROBAND	1024	1	Clinically normal; 1% of control RBC and lymphoblastoid cell APRT specific activity; 1% of control cross reacting material to anti-APRT antibodies in RBC's & lymphoblastoid cells; normal RBC HPRT activity; 46,XX
*GM7291		R1640 15I	0497	46	F	WH	--		AFF SIS	1024	2	Urolithiasis; onset at age 42; 1% of control RBC APRT specific activity; normal RBC HPRT activity; 1% of control cross-reacting material to anti-APRT antibodies in RBC
Adenosine Deaminase Deficiency With Immunodeficiency - 10270												
*GM2756	18HR	R1640 20I	0236	3	M	BL	--	A	PROBAND			Deficient ADA activity; enzyme phenotypes: 6PGD=A, G6PD=B, Peptidases A,C, & D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=3-2; < 1% of norm ADA act & cross immunoreactive protein in lymphoid cult; produces normal ADA mRNA
*GM2471	22HR	R1640 20I	0219		M	WH	--	A	PROBAND	219	1	Deficient RBC, fibroblast, and lymphoblast ADA activity; similarly affected female sib; enzyme phenotypes: G6PD=B, Neutral A-glucosidase C=1; see GM2434 Fibroblast; produces normal ADA mRNA
GM2472A	21HR	R1640 15I	0219		F	WH	+-(0)	A	MOTHER	219	2	Heterozygote level of ADA activity; enzyme phenotypes: Neutral A-glucosidase C =1, ADA=1; see GM2433 Fibroblast; two affected children

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency With Immunodeficiency - 10270												
*GM2606	23HR	R1640 20I	0223	8MO	M	WH	--	A	PROBAND	226	1	See GM2605 Fibroblast; deficient ADA activity; enzyme phenotypes: 6PGD=A, Peptidases A,C,& D=1; Acid A-glucosidase =1, Neutral A-glucosidase C=1; < 1% of norm ADA act & cross immunoreactive prot in lymph cult; produces norm ADA mRNA
*GM2608	16HR	R1640 20I	0223		F	WH	+-(0)	A	MOTHER	226	2	See GM2607 Fibroblast; enzyme phenotypes: Peptidases A,C,& D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=1; produces normal ADA mRNA
*GM2610	16HR	R1640 20I	0223		M	WH	+-(0)	A	FATHER	226	3	See GM2609 Fibroblast; enzyme phenotypes: Peptidases A,C,& D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=1; produces normal ADA mRNA
GM2581A		R1640 20I	0223	38	M	WH	+-(0)	A	FATHER	232	2	See GM2580 Fibroblast; enzyme phenotypes: Peptidases A,C,& D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=1; proband is GM2578 Fibroblast
GM2583	17HR	R1640 20I	0223	37	F	WH	+-(0)	A	MOTHER	232	3	See GM2582 Fibroblast; enzyme phenotypes: Peptidases A,C,& D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=1; proband is GM2578 Fibroblast
*GM1715	38HR	R1640 20I	0217	33MO	M	BL	--	A	PROBAND	423	1	Detroit 300; def. ADA activity; HLA type A2,30,B7,39; enzyme phenotypes: 6PGD=A, G6PD=A, Peptidases A,C,+D=1, Acid A-glucosidase=1, Neutral A-glucosidase C=3, & GLO-1=2; 46,XY; see GM2445 Fibroblast; produces normal ADA mRNA
*GM2825A	27HR	R1640 20I	0291	4MO	F	WH	--	A	PROBAND	446	1	Absent IgM and IgA; weak MLC response; absent T cells; see GM2824 Fibroblast; no detectable ADA activity in erythrocytes and lymphoblasts; possible structural defect in ADA mRNA or its precursors
*GM4258A		R1640 15I	0370	4	M	BL	--		PROBAND	694	1	Incompetent humoral and cellular immunity; less than 3% of normal ADA activity in erythrocytes, lymphocytes, and granulocytes; both parents have heterozygous ADA levels; produces normal ADA mRNA
Adenosine Deaminase Deficiency With No Immunodeficiency												
GM6142		R1640 15I	0208	7MO	M	WH	--	A	PROBAND			Santo Domingan; deficient ADA activity in RBCs; norm. act. in peripheral blood mononuclear cells & 65% in lymphoblasts; healthy (recurrent mild thrush); normal immunoglobulins; 80% of normal no. of T cells; normal response to mitogens

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency With No Immunodeficiency												
*GM7945		R1640 10U	0455	3MO	M	WH		A	PROBAND			Multiple malformations; failure to thrive; seizures; hemizygous range of Adenosine deaminase enzyme activity in RBC, fibroblasts, & lymphoid cell line; 46,XY,del(20)(pter>q13.1:)
*GM2294	21HR	R1640 20I	0208	10MO	M	BL	--	A	PROBAND	633	1	Normal serum levels of IgG, IgM, IgA, & IgE; deficient RBC ADA activity; partial ADA deficiency in fibro & lymphoblasts; see GM1997 Fibroblast; enzyme phenotypes 6PGD=A, G6PD=B, ADA=1, Neutral A-glucosidase C=2; produces normal ADA mRNA
*GM3043	28HR	R1640 20I	0170	16	M	BL	--	A	PROBAND	634	1	!Kung South African; see GM3029 Fibroblast; clinically normal; partial ADA deficiency; enzyme phenotypes: 6PGD=A, ADA=1, Neutral A-glucosidase C=2; contains unstable ADA protein and 3 to 4 times the norm level of norm ADA mRNA
*GM4396	21HR	R1640 15I	0251	6MO	M	WH	--	A	PROBAND	717	1	No detectable ADA activity in red cells or lymphocytes; see GM4395 Fibroblast; mother and brother have heterozygous levels of ADA activity; 4% of normal ADA activity and cross immunoreactive protein in lymphoid culture
GM5816		R1640 15I	0208	3WK	F	BL	--?	A	PROBAND	866	1	Clinically normal; adenosine deaminase deficiency in RBCs; partial deficiency in other tissues; possible variant subgroup; no thymic shadow by x-ray; 50% of normal purine nucleoside phosphorylase act.; 29% of norm. ADA act. in lympho.
GM5817		R1640 15I	0208	24	F	BL	+-(D)	A	MOTHER	866	2	Clinically normal; 50% of adenosine deaminase activity in RBCs; 50% of normal purine nucleoside phosphorylase activity; 91% of normal ADA activity in lymphoblasts
GM6200		R1640 15I	0208	3MO	F	BL	--	A	PROBAND	923	1	Clinically healthy except for recurrent thrush; not lymphopenic; partial ADA deficiency; normal mitogen responses, total lymphocytes, and % of T cells; 30% of normal ADA activity in lymphoblasts
GM6201A		R1640 15I	0208	20	F	BL	+-(D)		MOTHER	923	2	Clinically normal; <50% of normal RBC ADA enzyme activity

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Adenosine Deaminase Deficiency With No Immunodeficiency												
GM6143A		R1640 15I	0208	9MO	M	WH	--	A	PROBAND	961	1	Santo Domingan; deficient ADA activity in RBCs; normal activity in peripheral blood mononuclear cells; normal no. of T cells; normal response to mitogens; normal growth & development; similarly aff. sib; 86% of norm. ADA act. in lympho.
GM6415		R1640 15I	0208	32	M	WH	+-(O)	A	FATHER	961	2	Santo Domingan; clinically normal; two affected children; 71% of normal ADA activity in lymphoblasts
GM6416		R1640 15I	0208	30	F	WH	+-(O)	A	MOTHER	961	3	Santo Domingan; clinically normal; two affected children; 65% of normal ADA activity in lymphoblasts
GM7103		R1640 15I	0208	6WK	F	BL	--	A	PROBAND	996	1	Partial ADA deficiency; absent RBC ADA activity; 50% of normal lymphocyte ADA activity; normal growth and development; normal immunologic tests; 16% of normal ADA activity in lymphoblasts
GM7104		R1640 15I	0208	37	F	BL	+-(O)	A	MOTHER	996	2	Clinically normal; 17% of normal RBC ADA activity; 39% of normal ADA activity in lymphoblasts
Adenylate Kinase-1 Deficiency - 10300												
GM6052		R1640 15I	0009	7	F	BL			PROBAND	903	1	Clinically normal; no detectable AK activity in RBCs, 20% of normal activity in fibroblasts, and 17% of normal activity in leukocytes; similarly affected sib; parents have 50-60% of normal AK activity in RBCs
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency - 14752												
*GM1619	26HR	R1640 20I	0177	29	F	WH	+-	A	PROBAND	635	1	See GM1617 Fibroblast; deficient RBC ITPase; 20% of normal ITPase activity in lymphoblasts; Neutral Alpha-glucosidase C phenotype=1
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
*GM0467A		R1640 15I	0083		F			A	MUTANT			Chemically induced in vitro with E.M.S.; selectable with 8-azaguanine or 6-thioguanine
GM6804		R1640 15I	0066	16	M	WH	Y-		PROBAND	72	1	No detectable HPRT, normal APRT activity in fibroblasts; see GM1662 Fibroblast
*GM2292A	42HR	R1640 15I	0183	19FW	M	WH	Y-	A	PROBAND	74	1	Similarly affected brother; see GM2290 & GM2291 Fibroblast and GM2338 Amniotic; nonviable in HAT medium

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
GM1899A		R1640 15I	0066	10	M	WH	Y-	A	PROBAND	623	1	HLA type A2,A11,B18,B5(Bw35); nonviable in HAT medium
GM1900A		R1640 15I	0066		F	WH	+?		MOTHER	623	2	HLA type A2,A28,B18,B5(Bw35); no family history available; one affected son; 46, XX; 16% of cells show random chromosome loss/gain; clinically unaffected
GM7092		R1640 15I	0218	30MO	M	WH	Y-		PROBAND	990	1	Delayed motor development; choreoathetoid movements; elevated serum uric acid; no self-mutilation; deficient WBC HPRT activity, normal APRT activity; no males with neurologic disorders in pedigree
GM7093		R1640 15I	0218	32	F	WH	+-(O)		MOTHER	990	2	Carrier by hair root assay; one affected son; some family history of gout and renal stones
GM7094		R1640 15I	0218	34	M	WH	++		FATHER	990	3	Clinically normal; one affected son
Xeroderma Pigmentosum: Complementation Group A - 27870												
GM2344	21HR	R1640 20I	0173	42MO	M	WH	--		PROBAND	77	1	XP1WI; small for age; speech problems; deficient repair activity; see GM1630 Fibroblast
GM7689		R1640 15I	0441	37	M	WH	+-(O)		FATHER	77	2	See GM1631 Fibroblast; XPHF1WI; clinically unaffected
GM7690		R1640 15I	0441	33	F	WH	+-(O)		MOTHER	77	3	See GM1632 Fibroblast; XPHM1WI; clinically unaffected
*GM2250A		R1640 20I	0155	10	F	WH	--		PROBAND	484	1	XP12BE; same patient as GM2500; less than 2% of normal UV induced unscheduled DNA synthesis in fibroblasts; same patient as ATCC CRL 1223; see GM4429B SV40 Transformed Fibroblast and GM5509 Fibroblast
GM5511A		R1640 15I	0226	44	F	WH	+-(O)		MOTHER	484	2	Clinically normal; see GM5510 Fibroblast
GM5567		R1640 15I	0226	20	M	WH	+?		BRO	484	3	Clinically normal; possible heterozygote; see GM5566 Fibroblast
GM5569A		R1640 15I	0226	47	M	WH	+-(O)		FATHER	484	4	Clinically normal; see GM5568 Fibroblast
*GM2345A	26HR	R1640 15I	0211	8	F	OR	--		PROBAND	695	1	Japanese; XP2OS; severe XP; mental retardation; gait disturbance; see GM4312A SV40 Transformed Fibroblast; < 2% of normal UV induced unscheduled DNA synthesis in fibroblasts; similarly affected sister (GM4314A Fibroblast)

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM											
Xeroderma Pigmentosum: Complementation Group B - 27871											
*GM2252A		R1640 15I	0155	31	F	WH	--	PROBAND	283	1	Formerly GM1853; XP11BE; also affected with Cockayne syndrome; 3-7% of normal UV induced unscheduled DNA synthesis in fibroblasts; HLA type Aw32,w24,Bw40,B12
*GM1855	27HR	R1640 20I	0155	64	F	WH	+-(O)	MOTHER	283	2	See GM1854 Fibroblast; also heterozygous for Cockayne Syndrome; HLA type Aw32, (A1),B12,B27
Xeroderma Pigmentosum: Complementation Group C - 27872											
*GM2249B		R1640 15I	0155	16	M	WH	--	PROBAND	75	1	XP8BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; similarly affected twin; see GM0671 Fibroblast
*GM2498	42HR	R1640 20I	0155	16	M	WH	--	AFF TWIN	75	2	XP9BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; see GM0676 Fibroblast
*GM2246A	18HR	R1640 15I	0155	30	F	WH	--	PROBAND	482	1	XP1BE; 46,XX; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; same patient as GM2483
GM4238		R1640 20I	0226	59	M	WH	+-(O)	FATHER	482	2	Clinically normal; 46,XY; 12% of cells show random chromosome loss
GM4237A		R1640 15I	0226	56	F	WH	+-(O)	MOTHER	482	3	Clinically normal; 46,XX
GM4239		R1640 20I	0226	33	M	WH	+?	BRO	482	4	Possible heterozygote; clinically normal; 46,XY; 4% of cells show random chromosome loss
*GM2248A		R1640 15I	0155	25	M	WH	--	PROBAND	507	1	XP3BE; 46,XY; 10-20% of normal UV induced unscheduled DNA synthesis in fibroblasts; see GM0030A Fibroblast
*GM2634	40HR	R1640 20I	0226	16	F	BL	--	PROBAND	627	1	De Sanctis-Cacchione; lupus erythematosus; XP1MI; low level of UV induced unscheduled DNA synthesis in fibroblasts; see GM2096 Fibroblast
Xeroderma Pigmentosum: Complementation Group D - 27873											
*GM2485A		R1640 15I	0155	14	F	WH	--	PROBAND	624	1	XP7BE; 25-50% of normal UV induced unscheduled DNA synthesis in fibroblasts; same patient as GM2590; 46,XX
*GM2253C		R1640 15I	0155	14	M	BL	--	PROBAND	625	1	XP17BE; 25% of normal UV induced unscheduled DNA synthesis in fibroblasts; 46,XY; shows no increase above normals for chemical clastogen induced chromosome breakage

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM												
Xeroderma Pigmentosum: Complementation Group D - 27873												
*GM2473	36HR	R1640 20I	0155	34	F	BL	+-(D)		MOTHER	625	2	Clinically normal; 46,XX; 12% of cells show random chromosome loss
Xeroderma Pigmentosum: Complementation Group E - 27874												
*GM2450C		R1640 15I	0155	30	F	WH	--		AFF 2ND COU	626	2	XP3RO; 2nd cousin of XP2RO (GM2415 Fibroblast); 46,XX
Xeroderma Pigmentosum: Complementation Group H												
GM3249	21HR	R1640 20I	0110	5	M	WH	--		PROBAND	714	1	Also possible Cockayne Syndrome; XP skin changes in areas exposed to light; neurological manifestations; see GM3248 Fibroblast
Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878												
*GM2545	20HR	R1640 20I	0228	15	M	OR	--		PROBAND	499	1	Hawaiian; clinically affected
Xeroderma Pigmentosum: Variants - 27875												
*GM2449	34HR	R1640 20I	0110	30	M	WH	--		PROBAND			XPPHBE; variant type; 46,XY; 8% of cells show random chromosome loss, 2% show random gain
GM1646A		R1640 15I	0133	21	F	WH	--		PROBAND	377	1	See GM1389 Fibroblast; diagnosed at age 16 yrs; fibroblasts show normal UV sensitivity and repair but variant DNA replication post UV irradiation
Sun Sensitive High Cancer Syndrome												
GM6934		R1640 15I	0472	29	M	WH			PROBAND			Mental retardation, microcephaly, short stature, hypogonadism, photosensitivity, abnormal neurologic exam, and typical skin findings; no defects in DNA repair or replication observed in fibroblasts
DISORDERS OF STEROID METABOLISM												
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191												
GM2242	20HR	R1640 20I	0107	9MO	F	WH	--		PROBAND	85	1	See GM2241 Fibroblast; 21-hydroxylase deficiency; salt-losing type; responsive to hydrocortisone treatment
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010												
GM4873	20HR	R1640 15I	0175	11	M	WH	Y-	A	PROBAND	764	1	Elevated C26 & C22 long-chain fatty acid in cultured skin fibroblasts; adrenal insufficiency; progressive white matter disease; progressively deteriorating intellectual function; positive family history; see GM4904 Fibroblast

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DISORDERS OF STEROID METABOLISM												
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010												
GM4674	39HR	R1640 15I	0175	30	F	WH	+-(O)		MOTHER	764	2	Clinically normal; abnormal C26:C22 fatty acid ratio in plasma & cultured fibroblasts; 1 first cousin & 2 second cousins have adrenoleukodystrophy; see GM4903 Fibroblast
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Cockayne Syndrome - 21640												
*GM1712	43HR	R1640 20I	0006	21	M	WH	--		PROBAND	251	1	HLA type Aw32,A10,Bw15,B14,(A3); UV irradiation sensitive; see GM10988 Fibroblast; pt. is CS5HO
*GM1857	23HR	R1640 20I	0155	13	M	WH	--		PROBAND	399	1	HLA type: A9,A11;B7,Bw15; see GM1856 Fibroblast; fibroblasts show markedly decreased post U.V. light colony forming ability & same x-ray sensitivity as normal cells; pt. is CS3BE
GM2474A		R1640 15I	0155	29	M	WH	+-(O)		FATHER	628	2	Affected daughter is GM1629 Fibroblast
*GM2964	22HR	R1640 20I	0210	25	F	WH	--		PROBAND	636	1	Atypical; late age of onset; lack of ocular and retinal lesions; see GM2965 Fibroblast; lymphoid culture shows hypersensitivity to UV irradiation
Dystonia Musculorum Deformans, Dominant Form (Torsion Dystonia) - 12810												
GM2217	24HR	R1640 20I	0184	30	M	WH	+-		PROBAND	100	1	Age of onset 24 yrs; on Lithium and Valium treatment
GM2305	24HR	R1640 20I	0184	17	F	WH	+-		PROBAND	102	1	See GM2304 Fibroblast; age of onset 7 yrs; at time of venipuncture has very little speech and considerable dystonia of all extremities
GM2347	22HR	R1640 20I	0184	21	F	WH	+-		PROBAND	218	1	Swedish descent; age of onset 5 yrs
GM2264	19HR	R1640 20I	0184	14	F	WH	+-		PROBAND	475	1	Jewish; age of onset 8 yrs; minimal action dystonia; greatest difficulty with speech and chewing; parents are healthy
Dystonia Musculorum Deformans, Recessive Form (Torsion Dystonia) - 22450												
GM2307	35HR	R1640 20I	0184	13	M	WH	--		PROBAND	103	1	See GM2306 Fibroblast; age of onset 10 yrs; father is Jewish
GM2256	24HR	R1640 20I	0184	14	F		--		PROBAND	473	1	See GM2255 Fibroblast; age of onset 12 yrs; severe involvement of trunk and extremities; similarly affected sister

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Marfan Syndrome - 15470												
GM7727A		R1640 15I	0107	12	F	BL	+-		PROBAND	1070	1	Similarly affected mother; long face; high arched palate; genu valgum; mild scoliosis; tall; wide arm span; decreased U/L segment ratio; no chest or eye abnormalities; mitral valve prolapse; arachnodactyly; see GM7726 Fibroblast
Limb-Girdle Muscular Dystrophy (Pelvofemoral; Leyden-Moebius M. D.) - 25360												
GM3931A		R1640 15I	0348	67	M	WH	--		PROBAND	795	1	Myopathic EMG; elevated CPK; onset at age 18; slowly progressive proximal muscle weakness; impotence; deceased male sib had same disorder; see GM3932 Fibroblast
Muscular Dystrophy; Duchenne Type (Pseudohypertrophic Progressive) - 31020												
*GM6007		R1640 15I	0148	10	F		+-	A	PROBAND			Elevated CPK; EMG and muscle biopsy consistent with DMD; 46,X,t(X;9)(Xqter>Xp21.2::9p22>9pter;9qter>9p22::Xp21.2>Xpter); balanced; normal X is late replicating; parents both have normal karyotypes
GM3780	30HR	R1640 20I	0006	11	M	WH	Y-		PROBAND	533	1	Latin American; elevated serum CPK; similarly affected sib; see GM3781 Fibroblast
GM3782		R1640 20I	0006	10	M	WH	Y-		AFF BRO	533	2	Latin American; elevated serum CPK; see GM3783 Fibroblast
GM3929A		R1640 15I	0348	12	M	WH	Y-		PROBAND	824	1	Progressive weakness; pseudohypertrophy; similarly affected brother and male maternal cousins; see GM5162 Fibroblast
GM5159		R1640 15I	0348	47	F	WH	+-(D)		MOTHER	824	2	Clinically unaffected; 2 affected sons & 2 affected great nephews; see GM5158 Fibroblast
GM5161A		R1640 15I	0348	57	M	WH	++		FATHER	824	3	Clinically unaffected; 2 affected sons; see GM5160 Fibroblast
Myotonic Dystrophy (Steinert Disease) - 16090												
GM3094	28HR	R1640 20I	0210	27	M	WH	+-		PROBAND			Skeletal muscle myotonia; mild muscle weakness; abnormal glucose tolerance test
GM3756	25HR	R1640 20I	0348	44	M	WH	+-		PROBAND	488	1	Typical phenotype; mother, two brothers, and a sister also affected; see GM3755 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE											
Myotonic Dystrophy (Steinert Disease) - 16090											
GM3986A		R1640 15I	0348	39	M	WH	+-	AFF BRO	488	2	Distal weakness; clinical & electrical myotonia; cataracts; marked balding; see GM3987 Fibroblast
GM3990A		R1640 15I	0348	76	F	WH	+-	AFF MOTHER	488	4	Distal weakness; cataracts; clinical & electrical myotonia; frontal baldness; see GM3991 Fibroblast
GM3696A	28HR	R1640 20I	0348	12	F	WH	+-	AFF DAU	527	2	Family history of clinical phenotype; EMG myotonia and mild myopathic facies; affected father is GM3759 Fibroblast
GM3928		R1640 20I	0348	32	F	WH	++	SPOUSE	527	3	Clinically normal; mother of GM3696A; wife of GM3759 Fibroblast (proband)
GM3697A		R1640 15I	0348	17	M	WH	+-	PROBAND	590	1	Myotonic face; EMG myotonia; distal atrophy; positive family history
GM4034		R1640 15I	0348	48	M	WH	+-	AFF FATHER	590	2	Temporal atrophy; clinical myotonia; distal weakness; positive family history; see GM4033 Fibroblast
Osteopetrosis with Renal Tubular Acidosis - 25973											
*GM6066		R1640 15I	0126	32	F	WH	--	PROBAND	904	1	Diagnosed at age 3; mild anemia, multiple fractures, dull mentality, short stature, obese, decreased visual acuity, dental malocclusion, high arched palate, & mild genu valgum; 2 affected sisters; def. RBC carbonic anhydrase II activity
*GM6065		R1640 15I	0126	25	F	WH	--	AFF SIS	904	2	Diagnosed at age 1; short stature, multiple fractures, dull mentality, dental malocclusion, bilateral optic atrophy, high arched palate, and mild hirsutism; deficient RBC carbonic anhydrase II activity; see GM6071 Fibroblast
*GM6067		R1640 15I	0126	54	F	WH	+-(D)	MOTHER	904	3	Three affected daughters and one unaffected daughter; asymptomatic; 50% of normal RBC carbonic anhydrase II activity; slow growing culture; see GM6072 Fibroblast
*GM6068		R1640 15I	0126	56	M	WH	+-(D)	FATHER	904	4	Three affected daughters and one unaffected daughter; asymptomatic; 50% of normal RBC carbonic anhydrase II activity; see GM6073 Fibroblast

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF CONNECTIVE TISSUE, MUSCLE, AND BONE												
Osteopetrosis with Renal Tubular Acidosis - 25973												
*GM6087		R1640 15I	0126	30	F	WH	+-		SIS	904	6	Asymptomatic; 50% of normal RBC carbonic anhydrase II activity
*GM6119		R1640 15I	0126	46	F	WH	+-		PAT AUNT	904	9	Asymptomatic; 50% of normal RBC carbonic anhydrase II activity
GM7546		R1640 15I	0509	5	M	WH	--		PROBAND	1068	1	Puerto Rican; chronic acidosis, cerebral calcifications, poor weight gain, & mild developmental delay; parents are first cousins; deficient RBC carbonic anhydrase II activity
GM7547		R1640 15I	0509	43	F	WH	+-(O)		MOTHER	1068	2	Heterozygote level of RBC carbonic anhydrase II activity; clinically unaffected; one affected child
DISORDERS OF THE NERVOUS SYSTEM												
Apparently Normal Individuals With Normal Psychiatric Evaluations												
GM5377		R1640 15I	0308	24	F	WH			PROBAND			46,XX; 12% of cells show random chromosome loss
GM5380		R1640 15I	0308	32	F	WH			PROBAND			46,XX; 4% of cells show random chromosome loss
GM5392		R1640 15I	0308	27	F	WH			PROBAND			46,XX; 6% of cells show random chromosome loss
GM5398		R1640 15I	0308	44	M	WH			PROBAND			46,XY; 6% of cells show random chromosome loss
GM5408		R1640 15I	0308	28	F	WH			PROBAND			46,XX; 6% of cells show random chromosome loss/gain
GM5423		R1640 15I	0308	32	F	WH			PROBAND			46,XX; 4% of cells show random chromosome loss
GM6049		R1640 15I	0308	23	F	WH			PROBAND			46,XX; 16% of cells show random chromosome loss
GM6051		R1640 15I	0308	26	F	WH			PROBAND			46,XX; 6% of cells show random chromosome loss
GM6859A		R1640 15I	0308	25	F	WH			PROBAND			46,XX; 10% of cells show random chromosome loss
GM6862		R1640 15I	0308	34	F	WH			PROBAND			46,XX

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Apparently Normal Individuals With Normal Psychiatric Evaluations											
GM6867		R1640 15I	0308	34	F	WH		PROBAND			Hyperthymic personality; 46,XX; 12% of cells show random chromosome loss/gain
GM6875		R1640 15I	0308	26	M	WH		PROBAND			46,XY; 8% of cells show random chromosome loss/gain
GM6160		R1640 15I	0308	25	M	WH		PROBAND	1030	1	46,XY; 14% of cells show random chromosome loss/gain
GM7491		R1640 15I	0308	17	M	WH		PROBAND	1049	1	See GM7492 Fibroblast; 46,XY; 12% of cells show random chromosome loss/gain
GM7521		R1640 15I	0308	19	F	WH		PROBAND	1051	1	See GM7522 Fibroblast; 46,XX; 6% of cells show random chromosome loss
GM7544		R1640 15I	0308	22	F	WH		PROBAND	1055	1	See GM7545 Fibroblast; 46,XX; 4% of cells show random chromosome loss
GM7535		R1640 15I	0308	15	M	WH		PROBAND	1056	1	46,XY; 10% of cells show random chromosome loss
GM7533		R1640 15I	0308	16	F	OT		PROBAND	1057	1	Caucasian mother & Japanese father; see GM7532 Fibroblast; 46,XX; 14% of cells show random chromosome loss
GM7524		R1640 15I	0308	22	F	WH		PROBAND	1059	1	See GM7525 Fibroblast; 46,XX; 10% of cells show random chromosome loss
GM7752		R1640 15I	0308	17	M	WH		PROBAND	1071	1	See GM7753 Fibroblast; 46,XY; 12% of cells show random chromosome loss/gain
Dysautonomia (Riley-Day Syndrome) - 22390											
GM0599A		R1640 15I	0024		F	WH	--	PROBAND			Clinically affected
GM0600	24HR	R1640 20I	0024		M	WH	+-(0)	PARENT			Proband not in Repository; clinically unaffected
GM0601	90HR	R1640 20I	0024	10	M	WH	--	PROBAND			Clinically affected; 46,XY; 22% of cells show random chromosome loss
GM0602A	34HR	R1640 15I	0024		M	WH	+-(0)	PARENT			Proband not in Repository; clinically unaffected; 46,XY; 20% of cells show random chromosome loss/gain
GM0603	35HR	R1640 20I	0024		F	WH	+-(0)	PARENT			Proband not in Repository; clinically unaffected; 46,XX; 26% of cells show random chromosome loss

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Dysautonomia (Riley-Day Syndrome) - 22390												
GM1465	29HR	R1640 20I	0008	21	F	WH	--		PROBAND			Clinically affected
GM1466A		R1640 20I	0008	11	M	WH	--		PROBAND			Clinically affected
GM1777A		R1640 15I	0008	22	F	WH	--		PROBAND			Clinically affected
GM4635		R1640 15I	0110	53	F	WH	+-(O)		MOTHER	761	3	Clinically normal; Jewish (Ashkenazi); see GM4634 Fibroblast; proband is GM2343 Fibroblast
GM5041		R1640 15I	0191	23	M	WH	--		PROBAND	810	1	Jewish (Ashkenazi); severely affected; repeated respiratory and gastrointestinal crises; severe scoliosis; classical diagnostic features; absent papillae of tongue and abnormal histamine test; similarly affected sib
GM5042		R1640 15I	0191	18	F	WH	--		AFF SIS	810	2	Jewisn (Ashkenazi); severely affected; repeated respiratory & gastrointestinal crises; severe scoliosis; classical diagnostic features; absent papillae of tongue and abnormal histamine test
GM5043A		R1640 15I	0191	16	F	WH	+?		SIS	810	3	Possible heterozygote; normal histamine test; phenotypically normal
GM5044		R1640 15I	0191	22	F	WH	+?		SIS	810	4	Possible heterozygote; normal histamine test; phenotypically normal
GM5045A		R1640 15I	0191	59	M	WH	+-(O)		FATHER	810	5	Normal histamine test; phenotypically normal; two affected children
GM5046A		R1640 15I	0191	51	F	WH	+-(O)		MOTHER	810	6	Normal histamine test; phenotypically normal; two affected children
Friedreich Ataxia - 22930 + 13660												
GM3959		R1640 15I	0210	63	M	WH			PROBAND	68	1	Son and father also affected; pes cavus; peripheral neuropathy; cerebellar ataxia; nystagmus; sensorineural hearing loss; possibly dominant inheritance; see GM3295 Fibroblast
GM4079		R1640 15I	0348	30	M	WH			AFF BRO	594	2	Ataxia, cardiomyopathy, and mild peripheral neuropathy; see GM4078 Fibroblast; probably recessive inheritance; proband is GM3816 Fibroblast
Huntington's Disease - 14310												
*GM2080A	19HR	R1640 15I	0136	48	F	WH	+-		PROBAND	107	1	

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease - 14310												
GM2078	38HR	R1640 20I	0136	25	F	WH	+?		DAU	107	2	At risk (50%); mother and grandfather are affected; see GM2077 Fibroblast
*GM2146	22HR	R1640 20I	0186	54	M	WH	+-		PROBAND	108	1	Mother was also affected; see GM2147A Fibroblast; Neutral Alpha-glucosidase C phenotype=2-1
*GM2148	33HR	R1640 20I	0186	54	F	WH	++		SPOUSE	108	2	Normal wife; see GM2149 Fibroblast; Neutral Alpha-glucosidase C phenotype=1
*GM2150	20HR	R1640 20I	0186	26	F	WH	+?		DAU	108	3	At risk (50%); see GM2151 Fibroblast; personality disorder but no chorea; Neutral Alpha-glucosidase C phenotype= 2-1
GM2152A		R1640 15I	0186	40	F	WH	++		SPOUSE	109	2	Clinically normal widow of affected husband; proband not in Repository; see GM2153 Fibroblast
GM2154B		R1640 20I	0186	20	F	WH	+?		DAU	109	3	At risk (50%); father had H.D.; see GM2155 Fibroblast
GM2156	20HR	R1640 20I	0186	16	F	WH	+?		DAU	109	4	At risk (50%); father had H.D.; see GM2157 Fibroblast
GM2158	25HR	R1640 20I	0186	19	F	WH	+?		DAU	109	5	At risk (50%); father had H.D.; see GM2159 Fibroblast
GM2160	24HR	R1640 20I	0186	21	M	WH	+?		SON	109	6	At risk (50%); father had H.D.; see GM2161 Fibroblast
GM2162	21HR	R1640 20I	0186	12	M	WH	+?		SON	109	7	At risk (50%); father had H.D.; see GM2163 Fibroblast
GM2164B		R1640 15I	0186	58	M	WH	+-		PROBAND	110	1	Mother and sister were also affected; age of onset 46; see GM2165 Fibroblast; 46,XY
GM2176	34HR	R1640 20I	0186	26	M	WH	+?		SON	110	2	At risk (50%); see GM2177 Fibroblast; may show early chorea; has long standing personality problem
GM2182	25HR	R1640 20I	0186	21	F	WH	+?		DAU	110	3	At risk (50%); see GM2183 Fibroblast
GM2168A		R1640 20I	0186	52	M	WH	++		BRO-IN-LAW	110	4	Normal spouse of GM2166B; see GM2169 Fibroblast
GM2186	42HR	R1640 20I	0186	60	F	WH	+?		SIS	110	5	Asymptomatic at age 60; reduced at risk; see GM2187 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease - 14310											
GM2188	24HR	R1640 20I	0186	63	M	WH	++	BRO-IN-LAW	110	6	Normal spouse of GM2186; see GM2189 Fibroblast
GM2166B		R1640 20I	0186	58	F	WH	+-	AFF SIS	110	7	46,XX/47,XX,+12; (74%/26%); see GM2167A Fibroblast; mother and brother also have H.D.
GM2178		R1640 20I	0186	38	F	WH	+?	NIECE	110	8	Daughter of reduced at risk GM2186; reduced at risk
GM2180	29HR	R1640 20I	0186	51	F	WH	++	SPOUSE	110	9	Clinically unaffected wife of proband
GM3620		R1640 20I	0186	29	F	WH	+-	AFF DAU	110	10	See GM3621 Fibroblast; age of onset 18 years; similarly affected father and grandmother; 46,XX
*GM2172A	22HR	R1640 15I	0186	52	F	WH	+-	PROBAND	111	1	See GM2173A Fibroblast; Neutral Alpha - glucosidase C phenotype=1
GM2170	23HR	R1640 20I	0186	22	F	WH	+?	DAU	111	2	At risk (50%); see GM2171 Fibroblast
GM2174	22HR	R1640 20I	0186	55	M	WH	++	SPOUSE	111	3	Clinically normal; see GM2175 Fibroblast
GM2190		R1640 20I	0190	32	F	WH	+?	MOTHER	206	2	At risk (50%); mother of GM2123 Fibroblast; see also GM2191 Fibroblast
GM3642	31HR	R1640 15I	0136	55	M	WH	+-	PROBAND	292	1	See GM1061A Fibroblast; choreoathetosis, dysarthria, dysphagia; positive family history for H.D.
GM3644	24HR	R1640 15I	0136	48	M	WH	+-	PROBAND	293	1	See GM1085 Fibroblast; moderate ataxia, chorea, dysarthria, & dysphagia; onset at age 35; affected mother, grandmother, & sibs
GM3643	24HR	R1640 15I	0136	50	F	WH	+-	AFF SIS	293	2	See GM1083A Fibroblast; onset at age 35; choreoathetosis, mild dysarthria, & mild dysphagia
GM3645	20HR	R1640 20I	0136	53	F	WH	+-	AFF SIS	293	3	Severe choreoathetosis; moderate dysarthria and dysphagia; onset at age 33
GM3745		R1640 20I	0136	22	F	WH	+?	DAU	293	4	At risk (50%); father had H.D.
GM3746		R1640 20I	0136	24	F	WH	+?	DAU	293	5	At risk (50%); father had H.D.
GM3747		R1640 20I	0136	18	M	WH	+?	SON	293	6	At risk (50%); father had H.D.

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM									
Huntington's Disease - 14310												
GM3748		R1640 20I	0136	33	M	WH	+?		NEPHEW	293	7	Son of GM3645; at risk (50%); mother, aunt, and uncle have H.D.
GM3790		R1640 20I	0186	37	F	WH	+-		PROBAND	549	1	Chorea; bipolar affective disorder; dementia; 4 years duration
GM3794		R1640 20I	0186	19	F	WH	+?		DAU	549	3	At risk (50%); see GM3793 Fibroblast
GM3861		R1640 20I	0320	58	F	WH	+?		SIS	568	1	Reduced risk for H.D.; had ovarian cancer at age 50; see GM3860 Fibroblast
GM3871		R1640 20I	0320	56	F	WH	+?		SIS	568	2	Reduced at risk; also had malignant melanoma; see GM3870 Fibroblast
GM3865		R1640 20I	0320	51	F	WH	+-		PROBAND	568	3	See GM3864A Fibroblast; similarly affected sib
GM3873		R1640 20I	0320	31	M	WH	+?		SON	568	4	At risk (50%); mother had H.D.; see GM3872 Fibroblast
GM3867		R1640 20I	0320	26	F	WH	+?		DAU	568	5	At risk (50%); mother had H.D.; see GM3866 Fibroblast
GM3869		R1640 20I	0320	45	F	WH	+-		AFF SIS	568	6	See GM3868 Fibroblast; affected
GM3863		R1640 20I	0320	24	F	WH	+?		NIECE	568	7	At risk (50%); daughter of GM3869; see GM3862 Fibroblast
GM4017		R1640 15I	0136	56	F	WH	+-		PROBAND	587	1	Moderate to severe truncal & limb movements; mutism; diagnosed at age 49; father had H.D.
GM4021		R1640 15I	0136	20	F	WH	+?		DAU	587	3	At risk (50%); mother had H.D.
GM4023		R1640 15I	0136	28	F	WH	+?		DAU	587	4	At risk (50%); mother had H.D.; see GM4022 Fibroblast
*GM4219		R1640 20I	0367	78	M	WH	+-		PROBAND	682	1	Choreic movements and dementia; positive family history
*GM4189		R1640 20I	0367	77	F	WH	++		SPOUSE	682	2	Clinically normal; see GM4188 Fibroblast
*GM4197		R1640 20I	0367	51	F	WH	+-		AFF DAU	682	3	See GM4196 Fibroblast; affected
*GM4201A		R1640 15I	0367	53	M	WH	+-		AFF SON	682	5	See GM4200 Fibroblast; affected
*GM4191		R1640 20I	0367	85	F	WH	++		SIS-IN-LAW	682	6	Clinically normal; see GM4190 Fibroblast; mother of GM4195, GM4199 & GM4223

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease - 14310												
*GM4199		R1640 20I	0367	63	F	WH	+-		AFF NIECE	682	7	Daughter of GM4191; see GM4198 Fibroblast; father had H.D.
*GM4195		R1640 20I	0367	60	F	WH	+-		AFF NIECE	682	8	Daughter of GM4191; see GM4194 Fibroblast; father had H.D.
*GM4223		R1640 15I	0367	59	M	WH	+-		AFF NEPH	682	9	Son of GM4191; see GM4222 Fibroblast; father had H.D.
*GM4227		R1640 20I	0367	74	M	WH	+?		BRO	682	10	Reduced at risk; see GM4226 Fibroblast; mother had H.D.
*GM4233		R1640 15I	0367	70	M	WH	+?		BRO	682	11	Reduced at risk; see GM4232 Fibroblast; mother had H.D.
*GM4203		R1640 20I	0367	78	F	WH	+?		1ST COU	682	12	Reduced at risk; father had H.D.
*GM4193		R1640 20I	0367	71	M	WH	+?		1ST COU	682	13	Reduced at risk; mother had H.D.; see GM4192 Fibroblast
*GM4207		R1640 20I	0367	75	F	WH	+?		1ST COU	682	14	Reduced at risk; mother had H.D.; see GM4206 Fibroblast
*GM4205		R1640 20I	0367	81	M	WH	++		IN-LAW	682	15	Related by marriage; clinically normal; father of GM4211, 4213, 4231 & 4584; see GM4204 Fibroblast; wife had H.D.
*GM4217		R1640 20I	0367	40	F	WH	+-		AFF COU	682	16	Mother had H.D.; 1st cousin once removed to proband
*GM4229		R1640 20I	0367	52	F	WH	+-		AFF COU	682	17	Mother had H.D.; 1st cousin once removed to proband
*GM4225		R1640 20I	0367	47	M	WH	+?		COU	682	18	At risk (50%); mother had H.D.; 1st cousin once removed to proband
*GM4213		R1640 20I	0367	50	F	WH	+-		AFF COU	682	19	Early symptoms of H.D.; daughter of GM4205; see GM4212 Fibroblast; mother had H.D.; 1st cousin once removed to proband
*GM4231		R1640 15I	0367	55	M	WH	+-		AFF COU	682	20	Mother had H.D.; father is GM4205; daughter is GM4221; see GM4230 Fibroblast; 1st cousin once removed to proband
*GM4211		R1640 15I	0367	58	M	WH	+-		AFF COU	682	21	Father of GM4209; see GM4210 Fibroblast; mother had H.D.; 1st cousin once removed to proband

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease - 14310											
*GM4209		R1640 20I	0367	35	M	WH	+-	AFF COU	682	22	Son of GM4211; see GM4208 Fibroblast; 1st cousin twice removed to proband
*GM4221		R1640 20I	0367	31	F	WH	+?	COU	682	23	At risk (50%); daughter of GM4231; see GM4220 Fibroblast; 1st cousin twice removed to proband; father had H.D.
*GM4534	23HR	R1640 15I	0367	65	M	WH	+?	1ST COU	682	24	Reduced at risk; father had H.D.; sister is GM4203
*GM4584	36HR	R1640 15I	0367	50	M	WH	+?	COU	682	25	At risk (50%); mother had H.D.; son of GM4205; 1st cousin once removed to proband
*GM4894	27HR	R1640 15I	0367	60	M	WH	+-	AFF COU	682	26	First cousin once removed; choreic movements and dementia; see GM5031B Fibroblast
*GM4915A		R1640 15I	0367	56	M	WH	+-	AFF COU	682	27	Choreic movements and dementia; first cousin once removed to proband; see GM5030B Fibroblast
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4282		R1640 15I	0233	21	F	WH	--?	PROBAND	690	1	Code 48; rigid form of HD; possible homozygote; both parents affected; onset at age 14 yrs; neuro exam 3/82 shows a hypokinetic variant of HD with dystonia and marked tremor, ataxic wide-based gait; see GM4281 Fibroblast
*GM4866A		R1640 15I	0278	49	M	WH	+-	AFF FATHER	690	2	Code 47; clinically affected; neuro exam 3/81 shows rigidity and a narrow-based gait with pauses; see GM4867 Fibroblast
GM5736A		R1640 15I	0233		M	WH	+?	MAT UNCLE	690	3	Code 1173; unaffected; last examined on 4/82; father had HD; see GM5735Z Fibroblast
GM5726A		R1640 15I	0233	48	F	WH	+?	MAT AUNT	690	4	Code 1165; unaffected; last examined on 4/82; father had HD; see GM5725Z Fibroblast
GM6685		R1640 15I	0233		M	WH	+?	MAT UNCLE	690	5	Code 70; unaffected; last examined on 3/29/83; father had HD; affected spouse is GM4766A; affected daughter is GM6595; see GM6684Z Fibroblast
GM4772A		R1640 15I	0278	57	F	WH	+?	MAT AUNT	690	6	Code 441; unaffected; last examined on 3/27/81; two affected parents; spouse of HD patient; affected son is GM4284; see GM4773 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM4284	27HR	R1640 15I	0233	40	M	WH	+-		AFF 1ST COU	690	7	Code 442; mother is GM4772A; father had HD; neuro exam 3/82 shows prominent dyskinesia, fluid eye movements, facial grimacing, and problems swallowing; see GM4807 Fibroblast
GM4720A		R1640 15I	0278	39	F	WH	+?		MAT 1ST COU	690	8	Code 453; unaffected; last examined on 3/23/81; mother is GM4722A; father had HD; affected brother is GM4284; see GM4719 Fibroblast
GM4704A		R1640 15I	0278	35	F	WH	+?		MAT 1ST COU	690	9	Code 447; unaffected; last examined on 3/23/81; mother is GM4772A; father had HD; affected brother is GM4284; see GM4703 Fibroblast
GM4804A		R1640 15I	0278	34	F	WH	+?		MAT 1ST COU	690	10	Code 456; unaffected; last examined on 3/27/81; mother is GM4772A; father had HD; affected brother is GM4284; see GM4805 Fibroblast
GM5584A		R1640 15I	0233	23	M	WH	+?		MAT 1ST COU	690	11	Code 450; unaffected; last examined on 3/82; mother is GM4772A; father had HD; affected brother is GM4284; see GM5583Z Fibroblast
GM5590A		R1640 15I	0233	44	M	WH	+?		MAT COU	690	12	Code 483; unaffected; last examined on 3/82; stepfather and 1st cousin once removed to proband; brother is GM5580A; see GM5589Z Fibroblast
GM6370		R1640 15I	0233	30	F	WH	+?		MAT 2ND COU	690	13	Code 386; unaffected; last examined on 3/8/83; father had HD; see GM6369Z Fibroblast
GM5496A		R1640 15I	0278	22	M	WH	+?		MAT 2ND COU	690	15	Code 1195; unaffected; last examined on 3/15/82; father had HD; see GM5495Z Fibroblast
GM6769		R1640 15I	0233	21	F	WH	+?		MAT 2ND COU	690	16	Code 1196; unaffected; last examined on 4/4/83; father had HD; see GM6768Z Fibroblast
*GM5560B		R1640 15I	0233	38	M	WH	+-		AFF MAT COU	690	17	Code 619; clinically affected; affected sib is GM4776; 1st cousin once removed to proband; neuro exam 3/82 shows rigidity with reinforcement

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
*GM4776	29HR	R1640 15I	0233	53	M	WH	+-		AFF MAT COU	690	19	Code 608; clinically affected; expired (1983); affected son is GM4738A; see GM4777 Fibroblast
GM4820A		R1640 15I	0278	45	F	WH	++		MAT COU	690	20	Code 609; unaffected spouse of GM4776; affected son is GM4738A
GM4738A		R1640 15I	0278	20	M	WH	+-		AFF 2ND COU	690	21	Code 612; clinically affected; onset at age 18 yrs; affected father is GM4776; neuro exam.3/82 shows choreiform movements; see GM4737 Fibroblast
GM6384		R1640 15I	0233	30	F	WH	+?		MAT 2ND COU	690	22	Code 613; unaffected; last examined on 3/8/83; affected father is GM4776; affected brother is GM4738A; see GM6383Z Fibroblast
GM5638A		R1640 15I	0233	27	F	WH	+?		MAT 2ND COU	690	23	Code 614; unaffected; last examined on 3/82; affected father is GM4776; affected brother is GM4738A; see GM5637Z Fibroblast
GM5442		R1640 15I	0233	13	F	WH	+?		MAT 2ND COU	690	24	Code 615; unaffected; last examined on 3/15/82; affected father is GM4776; affected brother is GM4738A; see GM5441Z Fibroblast
GM5440B		R1640 15I	0233	18	F	WH	+?		MAT 2ND COU	690	25	Code 616; unaffected; last examined on 3/15/82; affected father is GM4776; affected brother is GM4738A; see GM5439Z Fibroblast
GM5506		R1640 15I	0233	18	F	WH	+?		MAT 2ND COU	690	26	Code 617; unaffected; last examined on 3/15/82; affected father is GM4776; affected brother is GM4738A; see GM5505Z Fibroblast
GM6366		R1640 15I	0233	61	M	WH	+?		MAT 1ST COU	690	27	Code 606; unaffected; last examined on 3/8/83; affected sibs are GM4776 and GM5560B; 1st cousin once removed to proband; see GM6365Z Fibroblast
GM6681		R1640 15I	0233	61	F	WH	+?		MAT COU	690	28	Code 1113; unaffected; last examined on 3/29/83; mother had HD; sib is GM5690A; see GM6680Z Fibroblast
GM4708A		R1640 15I	0278	49	F	WH	+?		MAT COU	690	29	Code 11; unaffected; last examined on 3/23/81; mother is GM5690A; spouse of GM4711 Fibroblast; see GM4707A Fibro.

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REF #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM5690A		R1640 15I	0233	60	F	WH	+?		MAT COU	690	43	Code 1083; unaffected; last examined on 4/82; sister of GM6681; mother of GM4708A; see GM5691Z Fibroblast
*GM6499		R1640 15I	0233	35	M	WH	+?		MAT COU	690	45	Code 188; unaffected; last examined on 3/14/83; affected sibs are GM4479, GM4876A, & GM4886A; see GM6498Z Fibro.
*GM4876A		R1640 15I	0278	34	F	WH	+-		AFF MAT COU	690	51	Code 189; clinically affected; onset at age 29 yrs; affected sibs are GM4886A & GM4479; see GM4877 Fibroblast
GM4864A		R1640 15I	0278	43	M	WH	+?		MAT COU	690	52	Code 190; unaffected; last examined on 4/4/81; spouse of GM4876A; mother is GM5558A; see GM4865 Fibroblast
*GM4718A		R1640 15I	0278	53	F	WH	+?		MAT COU	690	57	Code 119; unaffected; last examined on 3/23/81; affected sibs are GM4886A, GM4479, & GM4876A; unaffected spouse is GM6411Z Fibroblast; see GM4717 Fibro.
*GM4886A		R1640 15I	0278	48	F	WH	+-		AFF MAT COU	690	64	Code 184; clinically affected; affected sibs are GM4479 & GM4876A; see GM4887 Fibroblast
GM5530A		R1640 15I	0233	28	M	WH	+?		MAT COU	690	65	Code 1543; unaffected; last examined on 3/19/82; affected mother is GM4886A
GM5534A		R1640 15I	0233	22	M	WH	+?		MAT COU	690	69	Code 1561; unaffected; last examined on 3/19/82; affected mother is GM4886A; see GM5535Z Fibroblast
GM5544B		R1640 15I	0233	19	M	WH	+?		MAT COU	690	70	Code 1562; unaffected; last examined on 3/82; affected mother is GM4886A
GM4860A		R1640 15I	0278	19	M	WH	+?		MAT COU	690	71	Code 187; unaffected; last examined on 4/2/81; affected mother is GM4886A; see GM4861 Fibroblast
GM5526A		R1640 15I	0233	16	M	WH	+?		MAT COU	690	72	Code 1564; unaffected; last examined on 3/19/82; affected mother is GM4886A; see GM5527Z Fibroblast
GM5524A		R1640 15I	0233	14	M	WH	+?		MAT COU	690	74	Code 1566; unaffected; last examined on 3/19/82; affected mother is GM4886A
GM5528A		R1640 15I	0233	8	M	WH	+?		MAT COU	690	75	Code 1567; unaffected; last examined on 3/19/82; affected mother is GM4886A

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM5552		R1640 15I	0233	7	M	WH	+?		MAT COU	690	76	Code 1568; unaffected; last examined on 3/19/82; affected mother is GM4886A
*GM4479	33HR	R1640 15I	0233	52	F	WH	+-		AFF MAT COU	690	77	Code 9; clinically affected; affected sibs are GM4876A & GM4886A; affected spouse is GM4477; 2 affected children & 4 with probable HD; see GM4475 Fibro.
GM4477	62HR	R1640 15I	0233	58	M	WH	+-		AFF MAT COU	690	78	Code 8; clinically affected; onset at age 41 yrs; affected spouse is GM4479; 2 affected children & 4 with probable HD; neuro exam 3/82 shows difficulty in ambulation with frequent falls; see GM4476 Fibroblast
GM4844A		R1640 15I	0278	37	F	WH	+?		MAT COU	690	79	Code 134; clinically unaffected; aff. parents are GM4479 & GM4477; 2 aff. sibs & 4 with probable HD; unaff. spouse is GM6552Z Fib.; neuro exam 3/82 shows brisk jaw jerk, nystagmus, & rigidity; see GM4845 Fibroblast
GM4846A		R1640 15I	0278	31	M	WH	+?		MAT COU	690	89	Code 149; unaffected; last examined on 4/2/81; affected parents are GM4479 and GM4477; 2 affected sibs and 4 with probable HD; unaffected spouse is GM6553Z Fibroblast; see GM4847 Fibroblast
GM4690A		R1640 15I	0278	30	F	WH	+-?		MAT COU	690	94	Code 154; probable HD; last examined on 3/23/81; 2 affected sibs and 3 with probable HD; affected parents are GM4479 & GM4477; unaffected spouse is GM6467Z Fibroblast; see GM4689 Fibroblast
GM5542B		R1640 15I	0233	29	F	WH	+-		AFF MAT COU	691	7	Code 166; clinically affected; onset at age 24; affected parents are GM4479 & GM4477; 2 aff. sibs & 4 with probable HD; unaff. spouse is GM6728Z Fibro.; neuro exam 3/82 shows rigidity with reinforcement; see GM4849 Fibroblast
GM4289		R1640 15I	0233	26	F	WH	+?		MAT COU	691	9	Code 123; unaffected; last examined on 11/22/80; 2 affected sibs and 4 with probable HD; affected parents are GM4479 & GM4477; see GM4482 Fibroblast
GM4856A		R1640 15I	0278	24	F	WH	+-		AFF MAT COU	691	10	Code 170; clinically affected; onset at age 24 yrs; 1 affected sib and 4 with probable HD; affected parents are GM4479 & GM4477; neuro exam 3/82 shows bradykinesia; see GM4857 Fibroblast

REP ID	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6538		R1640 15I	0233		M	WH	++		IN-LAW	691	11	Code 171; unaffected spouse of GM4856A; see GM6539Z Fibroblast
GM6530		R1640 15I	0233	13	F	WH	+?		MAT COU	691	12	Code 1068; unaffected; last examined on 3/21/83; affected mother is GM4856A; see GM6531Z Fibroblast
GM6528		R1640 15I	0233	11	M	WH	+?		MAT COU	691	13	Code 1069; unaffected; last examined on 3/21/83; affected mother is GM4856A; see GM6529Z Fibroblast
GM6532		R1640 15I	0233	8	F	WH	+?		MAT COU	691	14	Code 1070; unaffected; last examined on 3/21/83; affected mother is GM4856A; see GM6533Z Fibroblast
GM4830A		R1640 15I	0278	23	M	WH	+-?		MAT COU	691	16	Code 172; probable HD; affected parents are GM4479 & GM4477; 2 affected sibs & 3 with probable HD; neuro exam 3/82 shows history of symptoms but no neurological signs; see GM4831 Fibroblast
GM5376		R1640 15I	0278	23	M	WH	+?		MAT COU	691	17	Code 173; unaffected; last examined on 2/3/82; affected parents are GM4479 and GM4477; 2 affected sibs and 4 with probable HD; see GM4837 Fibroblast
GM4828A		R1640 15I	0278	21	F	WH	+-?		MAT COU	691	18	Code 174; probable HD; affected parents are GM4479 & GM4477; 2 affected sibs & 3 with probable HD; unaffected spouse is GM6512Z Fibro.; neuro exam 3/82 shows mild left sided hyperreflexia & decreas. eyelash burying on rt; see GM4829 Fibro.
GM4858A		R1640 15I	0278	20	M	WH	+?		MAT COU	691	20	Code 178; unaffected; last examined on 4/2/81; affected parents are GM4479 and GM4477; 2 affected sibs and 4 with probable HD; see GM4859 Fibroblast
GM5536B		R1640 15I	0233	20	F	WH	+?		MAT COU	691	21	Code 179; unaffected; last examined on 3/21/83; affected parents are GM4479 and GM4477; 3 affected sibs & 4 with probable HD; see GM6541Z Fibroblast
GM4290	16HR	R1640 15I	0233	17	F	WH	+?		MAT COU	691	22	Code 180; unaffected; last examined on 3/19/82; affected parents are GM4477 and GM4479; 2 affected sibs and 4 with probable HD; see GM5551Z Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM4852A		R1640 15I	0278	15	M	WH	+?	MAT COU	691	23	Code 182; unaffected; last examined on 4/2/81; affected parents are GM4479 and GM4477; 2 affected sibs and 4 with probable HD; see GM4853 Fibroblast
GM4854A		R1640 15I	0278	12	M	WH	+?	MAT COU	691	24	Code 183; unaffected; last examined on 4/2/81; affected parents are GM4479 and GM4477; 2 affected sibs and 4 with probable HD; see GM4855 Fibroblast
GM6629		R1640 15I	0233	86	M	WH	+?	MAT GR UNC	691	25	Code 1040; unaffected; last examined on 3/83; probable escapee; mother had HD; see GM6630Z Fibroblast
GM5700A		R1640 15I	0233	61	F	WH	+?	PAT COU	691	26	Code 328; unaffected; last examined on 4/82; at risk father died at age 40; see GM5699Z Fibroblast
GM6421		R1640 15I	0233	56	M	WH	+?	PAT COU	691	27	Code 1483; unaffected; last examined on 3/11/83; at risk father died at age 40; see GM6422Z Fibroblast
GM5678A		R1640 15I	0233	58	M	WH	+-	AFF MAT COU	691	30	Code 356; clinically affected; affected sister is GM5610A; see GM5679Z Fibro.
GM5610A		R1640 15I	0278	52	F	WH	+-	AFF MAT COU	691	31	Code 344; clinically affected; affected brother is GM5678A; see GM5609Z Fibro.
GM4800A		R1640 15I	0278	45	F	WH	+?	MAT COU	691	32	Code 355; unaffected; last examined on 3/27/81; affected sibs are GM5678A & GM5610A; see GM4801 Fibroblast
GM4808A		R1640 15I	0278	42	M	WH	+?	MAT COU	691	33	Code 354; unaffected; last examined on 3/27/81; affected sibs are GM5678A & GM5610A; see GM4809 Fibroblast
GM5684B		R1640 15I	0278	72	F	WH	++	IN-LAW	691	35	Code 373; unaffected spouse of HD patient; affected children are GM5682A & GM5668A; see GM5685Z Fibroblast
GM5682A		R1640 15I	0278	49	F	WH	+-	AFF MAT COU	691	36	Code 374; clinically affected; father had HD; mother is GM5684B; affected brother is GM5668A; see GM5683Z Fibro.
GM5660A		R1640 15I	0233		F	WH	+?	MAT COU	691	37	Code 378; unaffected; last examined on 4/4/82; affected sibs are GM5682A and GM5668A; see GM5661Z Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM5668A		R1640 15I	0233	46	M	WH	+-		AFF MAT COU	691	38	Code 379; clinically affected; affected sister is GM5682A; see GM5669Z Fibro.
GM6376		R1640 15I	0233	57	F	WH	+?		MAT COU	691	39	Code 385; unaffected; last examined on 3/8/83; spouse of HD patient; mother of GM6370; see GM6375Z Fibroblast
GM6348		R1640 15I	0233	50	M	WH	+-		AFF MAT COU	691	40	Code 387; clinically affected; affected sibs are GM6443 & GM5554B; affected son is GM5626A; neuro exam 3/82 shows moderate chorea with parakinesias; see GM4819 Fibroblast
GM5642A		R1640 15I	0233	41	F	WH	++		COU	691	41	Code 388; unaffected spouse of GM6348; affected son is GM5626A; see GM5641Z Fibroblast
GM5626A		R1640 15I	0233	25	M	WH	+-		AFF MAT COU	691	42	Code 389; clinically affected; affected father is GM6348; see GM5625Z Fibroblast
GM6443		R1640 15I	0233	44	F	WH	+-		AFF MAT COU	691	43	Code 411; clinically affected; last examined 3/11/83; affected sibs are GM6348 and GM5554B; see GM6444Z Fibroblast
GM5554B		R1640 15I	0233		F	WH	+-		AFF MAT COU	691	44	Code 396; clinically affected; affected sibs are GM6348 & GM6443; see GM5555Z Fibroblast
GM5606A		R1640 15I	0233	37	F	WH	+?		MAT COU	691	46	Code 404; unaffected; last examined on 3/82; affected sibs are GM6348, GM6443, and GM5554B; see GM5605Z Fibroblast
GM4694	38HR	R1640 15I	0233	33	M	WH	+?		MAT COU	691	47	Code 414; unaffected; last examined on 3/23/81; affected sibs are GM6348, GM6443, and GM5554B; see GM4693 Fibro.
GM5600A		R1640 15I	0233	76	M	WH	++		IN-LAW	691	48	Code 419; unaffected spouse of HD patient; affected children are GM4798A, GM5622A, and GM5614A; see GM5599Z Fibro.
GM4796		R1640 15I	0233	50	F	WH	+?		MAT COU	691	49	Code 425; unaffected; last examined on 3/27/81; affected sibs are GM4798A, GM5622A, & GM5614A; see GM4797 Fibro.
GM4798A		R1640 15I	0278	48	M	WH	+-		AFF MAT COU	691	50	Code 420; clinically affected; affected sibs are GM5622A & GM5614A; neuro exam 3/82 shows flexion of distal interphalangeal joint of thumb & no vertical optokinetic response; see GM4799 Fibro.

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM5724A		R1640 15I	0233	46	M	WH	+?	MAT COU	691	51	Code 423; unaffected; last examined on 4/82; affected sibs are GM5622A, GM4798A, & GM5614A; see GM3723Z Fibro.
GM4794A		R1640 15I	0278	42	F	WH	+?	MAT COU	691	52	Code 426; unaffected; last examined on 3/27/81; affected sibs are GM4798A, GM5622A, & GM5614A; see GM4795 Fibro.
GM5622A		R1640 15I	0233	41	M	WH	+-	AFF MAT COU	691	53	Code 424; clinically affected; affected sibs are GM4798A & GM5614A; see GM5621Z Fibroblast
GM5614A		R1640 15I	0233	39	F	WH	+-	AFF MAT COU	691	54	Code 534; clinically affected; affected sibs are GM4798A & GM5622A; see GM5613Z Fibroblast
GM5740A		R1640 15I	0233	36	M	WH	+?	MAT COU	691	55	Code 1248; unaffected; last examined on 4/82; affected sibs are GM5622A, GM4798A, & GM5614A; see GM5739Z Fibro.
GM5640A		R1640 15I	0233	34	M	WH	+?	MAT COU	691	56	Code 1249; unaffected; last examined 3/82; affected sibs are GM5622A, GM4798A, & GM5614A; see GM5639Z Fibro.
GM4786A		R1640 15I	0278	54	F	WH	++	COU	691	58	Code 460; clinically unaffected spouse of HD patient; affected children are GM4716A, GM4710A, & GM4688A; see GM4787 Fibroblast
GM4716A		R1640 15I	0278	40	M	WH	+-	AFF MAT COU	691	59	Code 474; clinically affected; onset at age 19 yrs; affected sibs are GM4710A & GM4688A; neuro exam 3/82 shows slight chorea in toes, fingers, & shoulders; seems more demented than motor impaired; see GM4715 Fibroblast
GM4710A		R1640 15I	0278	38	F	WH	+-	AFF MAT COU	691	60	Code 463; clinically affected; onset at age 34 yrs; affected sibs are GM4716A & GM4688A; 1st cousin once removed to proband; neuro exam 3/82 shows dyskinesia & impaired gaze; see GM4709 Fibroblast
GM4688A		R1640 15I	0278	38	F	WH	+-	AFF MAT COU	691	61	Code 469; clinically affected; onset at age 34 yrs; affected sibs are GM4716A & GM4710A; neuro exam 3/82 shows mild chorea, marginal dystonia, hypomimia, and bradykinesia; see GM4687 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM5580A		R1640 15I	0233	45	M	WH	+?	MAT COU	691	63	Code 482; unaffected; last examined on 3/82; mother had HD; 1st cousin once removed to proband; see GM5579Z Fibroblast
GM5608A		R1640 15I	0233	54	F	WH	+-	AFF COU	691	64	Code 526; clinically affected; onset at age 47; both parents were affected; affected son is GM5628A; neuro exam 3/82 shows prominent chorea, moderate dystonia, & some dementia; see GM5607Z Fibro.
GM5594A		R1640 15I	0278	72	M	WH	++	IN-LAW	691	65	Code 527; unaffected spouse of GM5608A; affected son is GM5628A; see GM5593Z Fibroblast
GM5628A		R1640 15I	0233	36	M	WH	+-	AFF COU	691	66	Code 859; clinically affected; affected mother is GM5608A; see GM5627Z Fibro.
GM5546B		R1640 15I	0233	62	M	WH	+?	MAT COU	691	68	Code 7; unaffected; last examined on 3/82; 2 sibs had HD; paternal uncle of GM5608A; see GM5547Z Fibroblast
GM5688A		R1640 15I	0278	42	M	WH	+-	AFF 1ST COU	691	69	Code 1100; clinically affected; both parents were affected; affected sib is GM4868A; pat. cousin; see GM5689Z Fibro.
GM4868A		R1640 15I	0278	30	F	WH	+-	AFF PAT COU	691	70	Code 558; clinically affected; onset at age 29 yrs; both parents affected; affected sib is GM5688A; 1st cousin to proband; see GM4869 Fibroblast
GM6437		R1640 15I	0233	71	F	WH	+?	MAT COU	691	71	Code 67; unaffected; last examined on 3/11/83; one affected sib; spouse of HD patient; affected daughter is GM4766A; see GM6438Z Fibroblast
GM4766A		R1640 15I	0278	42	F	WH	+-	AFF MAT COU	691	72	Code 69; clinically affected; spouse is GM6685; affected daughter is GM6595; neuro exam 3/82 shows prominent chorea of all extremities & reduced saccades - vertical > horizontal; see GM4767 Fibro.
GM5604A		R1640 15I	0233	37	M	WH	+?	MAT COU	691	73	Code 72; unaffected; last examined on 3/82; affected sib is GM4766A; see GM5603Z Fibroblast
GM5572A		R1640 15I	0233	34	F	WH	+?	MAT COU	691	74	Code 73; unaffected; last examined on 3/82; affected sib is GM4766A; see GM5571Z Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM##	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6595		R1640 15I	0233	40	F	WH	+-?		1ST COU	691	76	Code 1158; probable HD; affected mother is GM4766A; father is GM6685; see GM6596Z Fibroblast
GM5558A		R1640 15I	0233	68	F	WH	+?		MAT COU	691	77	Code 555; unaffected; last examined on 3/82; one affected sib; son is GM4864A; see GM5559 Fibroblast
*GM4814A		R1640 15I	0278	74	M	WH	++		IN-LAW	691	78	Code 634; unaffected spouse of HD patient; affected children are GM4288 & GM4286; 4 other affected children are deceased; see GM4815 Fibroblast
*GM4792A		R1640 15I	0278	52	M	WH	+?		MAT COU	691	79	Code 640; unaffected; last examined on 3/27/81; affected sibs are GM4288 and GM4286; 4 others are deceased; father is GM4814A; see GM4793 Fibroblast
*GM4288	15HR	R1640 15I	0233	44	M	WH	+-		AFF MAT COU	691	81	Code 647; clinically affected; onset at age 30 yrs; affected sib is GM4286; affected dau is GM4724; father is GM4814A; neuro exam 3/82 shows bradykinesis, Parkinsonian features, mild chorea, PD gait & dystonia; see GM4287 Fibroblast
*GM4724	32HR	R1640 15I	0233	19	F	WH	+-		AFF MAT COU	691	82	Code 653; clinically affected; affected father is GM4288; neuro exam 3/82 shows bradykinesis, juvenile features of HD with markedly reduced saccadic velocity, Parkinsonian features, and marginal dystonia; see GM4723 Fibroblast
*GM4286		R1640 15I	0233	41	M	WH	+-		AFF MAT COU	691	83	Code 659; clinically affected; onset at age 30 yrs; affected sib is GM4288; 4 others are deceased; father is GM4814A; neuro exam 3/82 shows bradykinesis, dystonia, & chorea; see GM4285 Fibroblast
*GM4730A		R1640 15I	0278	39	F	WH	+?		MAT COU	691	84	Code 665; unaffected; last examined on 3/27/81; affected sibs are GM4288 and GM4286; 4 others are deceased; father is GM4814A; see GM4729 Fibroblast
*GM4790A		R1640 15I	0278	101	F	WH	+?		COU	691	85	Code 663; unaffected; last examined on 3/27/81; probable escapee; spouse of HD patient; see GM4791 Fibroblast
*GM4736A		R1640 15I	0278	62	M	WH	+?		MAT COU	691	86	Code 560; unaffected; last examined on 3/23/81; son of GM4790A; affected spouse is GM4818B; affected daughter is GM4722A; see GM4735 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
*GM4818B		R1640 15I	0233	60	F	WH	+-	AFF PAT AUNT	691	87	Code 559; clinically affected; spouse is GM4736A; affected daughter is GM4722A; neuro exam 3/82 shows inability to talk, ability to walk only 4-5 steps with assistance; terminal stages of HD; see GM5562Z Fibroblast
GM4722A		R1640 15I	0278	37	F	WH	+-	AFF PAT COU	691	88	Code 561; clinically affected; parents are GM4736A & GM4818B; neuro exam 3/82 shows chorea & facial twitches; 1st cousin to proband; see GM4721 Fibroblast
GM6777		R1640 15I	0233	76	M	WH	+?	MAT COU	691	89	Code 917; unaffected; last examined on 4/4/83; affected sib is GM6767; father had HD; see GM6776Z Fibroblast
GM6633		R1640 15I	0233	51	M	WH	+?	MAT COU	691	90	Code 920; unaffected; last examined on 3/26/83; affected sib is GM6767; see GM6634Z Fibroblast
GM6655A		R1640 15I	0233	63	F	WH	+?	MAT COU	691	91	Code 923; unaffected; last examined on 3/83; affected sib is GM6767; see GM6656Z Fibroblast
GM6767		R1640 15I	0233	61	M	WH	+-	AFF MAT COU	691	92	Code 929; clinically affected; affected child is GM6651A; unaffected spouse is GM6615Z Fibroblast; see GM6766Z Fibro.
GM6651A		R1640 15I	0233	45	M	WH	+-?	MAT COU	691	94	Code 931; probable HD; affected father is GM6767; see GM6652Z Fibroblast
GM6631		R1640 15I	0233	41	M	WH	+-	AFF MAT COU	691	95	Code 934; clinically affected; affected sib is GM6651; affected father is GM6767; see GM6632Z Fibroblast
GM6593A		R1640 15I	0233	55	M	WH	+?	MAT COU	691	96	Code 940; unaffected; last examined on 3/24/83; affected sibs are GM5428, GM6566, and GM6447; see GM6594Z Fibro.
GM5428		R1640 15I	0233	44	F	WH	+-	AFF MAT COU	691	98	Code 963; clinically affected; affected sibs are GM6566 and GM6447; see GM5427Z Fibroblast
GM6585A		R1640 15I	0233	43	M	WH	+?	MAT COU	691	99	Code 994; unaffected; last examined on 3/83; affected sibs are GM5428, GM6566, & GM6447; see GM6586Z Fibroblast
GM6566		R1640 15I	0233	37	F	WH	+-	AFF MAT COU	692	1	Code 1002; clinically affected; affected sibs are GM5428 and GM6447; see GM6567Z Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM								
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)												
GM6447		R1640 15I	0233	37	F	WH	+-?		MAT COU	692	2	Code 932; probable HD; affected sibs are GM5428 and GM6566; affected spouse is GM6651A; see GM6448Z Fibroblast
GM6423		R1640 15I	0233	19	F	WH	+?		MAT COU	692	3	Code 2992; unaffected; last examined on 3/11/83; affected parents are GM6447 & GM6651A; see GM6424Z Fibroblast
GM6449		R1640 15I	0233	16	F	WH	+?		MAT COU	692	4	Code 2996; unaffected; last examined on 3/11/83; affected parents are GM6447 & GM6651A; see GM6450Z Fibroblast
GM6445		R1640 15I	0233	14	F	WH	+?		MAT COU	692	5	Code 2999; unaffected; last examined on 3/11/83; affected parents are GM6447 & GM6651A; see GM6446Z Fibroblast
GM6601		R1640 15I	0233	18	F	WH	+?		MAT COU	692	6	Code 2993; unaffected; last examined on 3/24/83; affected parents are GM6447 & GM6651A; see GM6602Z Fibroblast
GM6643		R1640 15I	0233	23	M	WH	+?		MAT COU	692	7	Code 933; unaffected; last examined on 3/26/83; affected parents are GM6447 & GM6651A; see GM6644Z Fibroblast
GM5486A		R1640 15I	0233	33	M	WH	+?		MAT COU	692	8	Code 1010; unaffected; last examined on 3/82; affected sibs are GM5428, GM6566, & GM6447; see GM5485Z Fibroblast
GM6581		R1640 15I	0233	29	F	WH	+-		AFF COU	692	9	Code 950; clinically affected; niece of GM5428; see GM6582Z Fibroblast
GM5448		R1640 15I	0233	46	M	WH	+?		PAT 1ST COU	692	10	Code 533; unaffected; last examined on 3/15/82; affected spouse is GM5614A; 1st cousin once removed to proband; see GM5447Z Fibroblast
*GM5452A		R1640 15I	0233	44	F	WH	+?		PAT 1ST COU	692	11	Code 537; unaffected; last examined on 3/15/82; 1st cousin once removed to proband; see GM5451Z Fibroblast
*GM5454A		R1640 15I	0233	38	F	WH	+?		PAT 1ST COU	692	12	Code 543; unaffected; last examined on 3/82; 1st cousin once removed to proband; see GM5453Z Fibroblast
GM5476A		R1640 15I	0233	35	M	WH	+?		PAT 1ST COU	692	13	Code 547; unaffected; last examined on 3/11/83; 1st cousin once removed to proband; see GM5475Z Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
Huntington's Disease: Venezuelan Family - 14310 (See Appendix for Pedigree)											
GM6481		R1640 15I	0233	72	M	WH	+?	PAT UNCLE	692	14	Code 552; unaffected; last examined on 3/14/83; 5 affected sibs; brother of GM4866A; see GM6480Z Fibroblast
*GM6364		R1640 15I	0233	59	F	WH	+?	PAT AUNT	692	15	Code 585; unaffected; last examined on 3/8/83; 5 affected sibs; brother is GM4866A; see GM6363Z Fibroblast
*GM4734A		R1640 15I	0278	57	F	WH	+?	PAT AUNT	692	16	Code 586; unaffected; last examined on 3/23/81; 5 affected sibs; brother is GM4866A; see GM4745 Fibroblast
*GM4692A		R1640 15I	0278	31	M	WH	+-	AFF 1ST COU	692	19	Code 589; clinically affected; onset at age 25 years; affected sib is GM6439; neuro exam 3/82 shows prominent laughing (? pseudo-bulba) & involuntary vocalizations, dementia, & dystonia; see GM4691 Fibroblast
*GM6439		R1640 15I	0233	29	M	WH	+-	AFF 1ST COU	692	20	Code 591; clinically affected; affected sib is GM4692A; neuro exam 3/82 shows reduced vertical > horizontal saccades, mild dystonia, & prominent chorea; see GM6440Z Fibroblast
GM6773		R1640 15I	0233	37	M	WH	+-	AFF PAT UNC	692	21	Code 598; clinically affected; last examined on 4/4/83; four affected sibs; brother of GM4866A; see GM6772Z Fibro.
GM6677		R1640 15I	0233	47	M	WH	+?	PAT UNCLE	692	22	Code 603; unaffected; last examined on 3/29/83; 5 affected sibs; brother is GM4866A; see GM6676Z Fibroblast
*GM4780A		R1640 15I	0278	71	M	WH	+?	PAT GR UNC	692	23	Code 624; unaffected; last examined on 3/27/81; 4 affected sibs; see GM4781 Fibroblast
Olivopontocerebellar Atrophy - 16440, 50, 60, 70, and 25830											
GM3562		R1640 20I	0110	22	F	BL		PROBAND	240	1	Probably type III; see GM3561 Fibroblast; positive family history
*GM6926		R1640 15I	0473	29	M	WH	+-	PROBAND	972	1	Olivopontocerebellar atrophy type I; spinocerebellar ataxia; six affected generations; see GM6927 Fibroblast
Primary Affective Disorder - 12548 and 30920											
GM6876		R1640 15I	0308	49	F	WH		PROBAND			Type II bipolar illness with major depression; hyperthymic personality; onset at age 15; numerous episodes of major depression; 2 episodes of hypomania; hospitalized once; received ECT, Parnate, & Stelazine

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Primary Affective Disorder - 12548 and 30920												
GM7097		R1640 15I	0308	50	F	WH			PROBAND			Type I bipolar illness; onset at age 8; multiple episodes of depression, mania, & panic attacks; has received Elavil, Stelazine, Triavil, & Tofranil; no hospitalizations
GM6138		R1640 15I	0308	49	M	WH			FATHER	802	3	Normal psychiatric evaluation; type I bipolar daughter is GM5215 Fibroblast
GM6139		R1640 15I	0308	44	F	WH			AFF MOTHER	802	4	Depressive personality; onset at age 13 yrs; premenstrual depression and irritability; type I bipolar daughter is GM5215 Fibroblast
*GM4984		R1640 15I	0308	20	M	WH			PROBAND	811	1	Type I bipolar illness; onset at age 17; three manic episodes; drug abuse; on Lithium; positive family history; see GM5224 Fibroblast
*GM4986		R1640 15I	0308	28	M	WH			AFF BRO	811	2	Type I bipolar illness; onset at age 17; 5 major depressive episodes and 2 manic episodes; on Lithium; see GM5225 Fibro.
*GM5909A		R1640 15I	0308	28	M	WH			BRO	811	3	Clinically normal; two sibs with type I bipolar illness
*GM5946A		R1640 15I	0308	62	M	WH			FATHER	811	4	Clinically unaffected; two children have type I bipolar illness; twin sister has unipolar illness
*GM5945		R1640 15I	0308	55	F	WH			MOTHER	811	5	Clinically unaffected; two children with type I bipolar illness; sister has type II bipolar illness; mother had unipolar illness
*GM5910		R1640 15I	0308	25	F	WH			AFF SIS	811	6	Two sibs with type I bipolar illness; minor depression; drug abuse; sexual deviation
*GM7200		R1640 15I	0308	16	M	WH			AFF MAT COU	811	7	Cyclothymic personality; minor depression; onset at age 14; see GM7199 Fibroblast; maternal first cousin of a bipolar male
*GM7208		R1640 15I	0308	28	F	WH			AFF MAT COU	811	8	One episode of hypomania; onset at age 25; no hospitalization; no medication; maternal first cousin of a bipolar male; see GM7209 Fibroblast; sister of GM7200

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
*GM7210		R1640 15I	0308	26	F	WH		MAT COU	811	9	Clinically unaffected; see GM7211 Fibroblast; sister of GM7200; maternal first cousin of a bipolar male
*GM7268		R1640 15I	0308	30	F	WH		MAT COU	811	10	At risk; clinically unaffected; see GM7269 Fibroblast; maternal first cousin of a bipolar male; sister of GM7200; paternal aunt & cousin have bipolar illness
GM7392		R1640 15I	0308	48	F	WH		AFF MAT AUNT	811	11	Type II bipolar illness with major depression; onset at age 34; 2 episodes of major depression; 1 hypomanic episode; hospitalized; on Lithium and Elavil; see GM7393 Fibroblast
GM7411		R1640 15I	0308	59	F	WH		AUNT	811	12	Clinically unaffected; in-law of proband; mother of GM7200, GM7208, GM7210, and GM7268; wife of GM7423; see GM7413 Fibroblast
GM7423		R1640 15I	0308	63	M	WH		MAT UNCLE	811	13	Clinically unaffected; reduced at risk; husband of GM7411; father of GM7200, GM7208, GM7210, and GM7268; see GM7424 Fibroblast
GM4985		R1640 15I	0308	43	F	WH		PROBAND	812	1	Type I bipolar illness; positive family history; onset at age 18; 3 episodes of major depression; indeterminate number of manic episodes; on Lithium; see GM5226 Fibroblast
GM4946A		R1640 15I	0308	55	M	WH		AFF BRO	812	2	Schizo-affective manic and schizo-affective depressed; anti-social personality; onset at age 19; 4 episodes of major depression, one episode of mania, and 4 episodes of hypomania; on Lithium; see GM5218A Fibroblast
GM6031		R1640 15I	0308	61	M	WH		AFF BRO	812	3	Cyclothymic personality; generalized anxiety disorder; onset at age 49
GM5947		R1640 15I	0308	27	F	WH		AFF NIECE	812	4	Minor depression; bulimia; generalized anxiety disorder; Cushing's disease; onset at age 23; two episodes of minor depression; on Nitotane and Hydrochlorathiazide

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMB	FAMM	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Primary Affective Disorder - 12548 and 30920												
GM6140		R1640 15I	0308	54	F	WH			SIS	812	5	Normal psychiatric evaluation; three sibs with primary affective disorders
GM7378		R1640 15I	0308	15	F	WH			DAU	812	6	At risk; mother has type I bipolar illness; see GM7379 Fibroblast
GM5134	Z5HR	R1640 15I	0308	24	M	WH			PROBAND	820	1	Type I bipolar illness; positive family history; onset at age 15; one major depressive episode; two manic episodes; on Lithium
GM5135		R1640 15I	0308	47	F	WH			AFF MOTHER	820	2	Type I bipolar illness; positive family history; onset at age 21; four major depressive episodes; one manic episode; on Lithium
GM7239		R1640 15I	0308	21	F	WH			SIS	820	3	At risk; clinically unaffected; mother and brother have type I bipolar illness; see GM7240 Fibroblast
GM7245		R1640 15I	0308	24	M	WH			AFF BRO	820	4	Alcoholism; minor depression; onset of alcoholism at age 15 and depression at age 23; mother and brother have type I bipolar illness; see GM7246 Fibroblast
GM5156	30HR	R1640 15I	0308	47	F	WH			PROBAND	823	1	Type I bipolar illness; positive family history; onset at age 31; two episodes of major depression; four episodes of mania; on Lithium; two similarly affected sons and one daughter
GM6972		R1640 15I	0308	20	F	WH			DAU	823	3	At risk; clinically unaffected; mother, sister, & 2 brothers have type I bipolar illness; see GM7398 Fibroblast
GM6973		R1640 15I	0308	50	M	WH			SPOUSE	823	4	Clinically unaffected; wife, daughter, & 2 sons have type I bipolar illness; see GM7321 Lymphoid
GM6974		R1640 15I	0308	18	F	WH			DAU	823	5	At risk; clinically unaffected; mother, sister, & 2 brothers have type I bipolar illness; see GM7320 Fibroblast
GM7218		R1640 15I	0308	28	F	WH			AFF DAU	823	6	Type I bipolar illness; hyperthymic personality; generalized anxiety; multiple episodes of depression; 3 episodes of mania; currently receiving Lithium, Haldol, Cogentin, & Thorazine; see GM7219 Fibroblast

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Primary Affective Disorder - 12548 and 30920												
GM7316		R1640 15I	0308	53	F	WH			SIS	823	7	Clinically unaffected; at risk; sister has type I bipolar illness; see GM7317 Fibroblast
GM7326		R1640 15I	0308	56	M	WH			BRO	823	8	Clinically unaffected; sister has type I bipolar illness
GM7335A		R1640 15I	0308	56	M	WH			BRO	823	9	Clinically unaffected; one episode of minor depression after receiving Butazolidine for arthritis; sister has bipolar manic-depressive illness; see GM7336 Fibroblast
GM7394		R1640 15I	0308	26	M	WH			AFF SON	823	10	Type I bipolar illness; drug abuse; generalized anxiety disorder; anti-social behavior; onset at age 13; one manic episode lasting 6 weeks; hospitalized; on Haldol and Lithium; see GM7395 Fibroblast
GM7550		R1640 15I	0308	40	F	WH			SIS	823	11	At risk; clinically unaffected; sibling of a type I bipolar female; see GM7551 Fibroblast
GM4942A		R1640 15I	0308	58	F	WH			PROBAND	829	1	Type I bipolar illness; positive family history; onset at age 33; one manic episode and 3 major depressive episodes; on Lithium
GM4972	41HR	R1640 15I	0308	60	F	WH			PROBAND	830	1	Unipolar illness; positive family history; two episodes of dysphoria; poor appetite; loss of energy, loss of interest, & slowed down movements; onset at age 49; on Elavil; see GM5216 Fibroblast
GM7152		R1640 15I	0308	27	M	WH			AFF NEPHEW	830	2	Probable unipolar illness; major depression; onset at age 17; required hospitalization & medication; son of GM7154; see GM7153 Fibroblast
GM7154		R1640 15I	0308	65	M	WH			AFF BRO	830	3	Type I bipolar illness; onset at age 18; indeterminate number of episodes of major depression; indeterminate number of manic episodes; no hospitalizations; no medication; see GM7155 Fibroblast
GM7201		R1640 15I	0308	25	F	WH			AFF DAU	830	4	Type II bipolar illness; major depression; onset at age 15; 2 episodes of depression; 2 episodes of hypomania; treated with Elavil and Lithium; see GM7209 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
			Primary Affective Disorder - 12548 and 30920								
GM7220		R1640 15I	0308	32	F	WH		DAU	830	5	At risk; see GM7221 Fibroblast; mother has unipolar illness, 1 sister has type II bipolar illness, & 1 sister has minor depression
GM7222		R1640 15I	0308	30	F	WH		NIECE	830	6	At risk; father (GM7154) has type I bipolar illness; see GM7223B Fibroblast
GM7257		R1640 15I	0308	29	F	WH		AFF NIECE	830	7	Type I bipolar illness; onset at age 17; at least 10 episodes of major depression; 2 episodes of mania; hospitalizations; on Lithium, Mellaril, Haldol, Cogentin, Elavil, Thorazine, & Stelazine; see GM7258 Fibroblast
GM7324		R1640 15I	0308	60	F	WH		AFF SIS	830	8	Type I bipolar illness; mother of GM7257; see GM7325 Fibroblast
GM7328		R1640 15I	0308	72	M	WH		BRO	830	9	Clinically unaffected; brother and sister have bipolar illness; see GM7329 Fibroblast
GM7330		R1640 15I	0308	63	M	WH		BRO-IN-LAW	830	10	Alcoholism; husband of GM7324; father of GM7257; see GM7331 Fibroblast
GM7748		R1640 15I	0308	36	M	WH		SON	830	11	At risk; hyperthymic personality onset at age 13; mother has unipolar illness & a sister has type II bipolar illness; see GM7749 Fibroblast
GM7800		R1640 15I	0308	62	M	WH		SPOUSE	830	12	Clinically unaffected; wife has unipolar illness; one daughter has type II bipolar illness and another minor depression; see GM7924 Fibroblast
GM4996		R1640 15I	0308	27	F	WH		PROBAND	832	2	Type I bipolar illness; positive family history; onset at age 22; 3 episodes of major depression; indeterminate number of episodes of mania; hyperthymic personality; on Lithium; see GM5227 Fibro.
GM5921		R1640 15I	0308	66	M	WH		AFF FATHER	832	3	One major depressive episode with hospitalization; onset at age 61; also has diabetes mellitus since age 47
GM5922		R1640 15I	0308	61	F	WH		AFF MOTHER	832	4	Two episodes of major depression; onset at age 54

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMD	FAMM	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
GM6022		R1640 15I	0308	31	F	WH		AFF SIS	832	5	Cyclothymic personality; grief reaction-minor depression; onset at age 12
GM6032		R1640 15I	0308	26	F	WH		AFF SIS	832	6	Depressive personality; anti-social personality; minor depression; drug abuse; alcoholism
GM4949B		R1640 15I	0308	55	M	WH		PROBAND	833	1	Type I bipolar illness; positive family history; onset at age 38; three episodes of major depression; five episodes of mania; on Lithium; see GM5220 Fibroblast
GM4952A		R1640 15I	0308	65	M	WH		PROBAND	834	1	Type II bipolar illness; positive family history; onset at age 43; three episodes of major depression; three episodes of hypomania; on Lithium; a daughter has acute intermittent porphyria; patient tested negative; see GM5221 Fibroblast
GM4950A		R1640 15I	0308	38	M	WH		PROBAND	835	1	Type I bipolar illness; positive family history; onset at age 32; hyperthymic personality; four episodes of mania; unknown number of minor depressive episodes; on Lithium; see GM5222 Fibroblast
GM7241		R1640 15I	0308	18	M	WH		PROBAND	836	1	See GM7242 Fibroblast; at risk; mother has type II bipolar illness
GM7249		R1640 15I	0308	18	M	WH		TWIN	836	3	See GM7250 Fibroblast; at risk; dizygotic twin of GM7241; mother has type II bipolar illness
GM5240		R1640 15I	0308	46	F	WH		PROBAND	847	1	Type II bipolar illness with major depression; positive family history; onset at age 20; indeterminate number of major depressive episodes; 10 hypomanic episodes
GM5237B		R1640 15I	0308	49	M	WH		SPOUSE	847	2	One year history of alcoholism at age 45; has never received psychiatric treatment
GM5236		R1640 15I	0308	21	M	WH		AFF SON	847	3	Type II bipolar illness; onset at age 18; two episodes of minor depression and one episode of major depression
GM5239		R1640 15I	0308	15	F	WH		AFF DAU	847	4	Type II bipolar illness; onset at age 15; one episode of major depression; one episode of minor depression; and one hypomanic episode

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
GM5651A		R1640 15I	0308		M	WH		AFF SON	847	6	Type II bipolar illness; minor depression
GM5883		R1640 15I	0308	39	F	WH		PROBAND	888	1	Type II bipolar illness; onset at age 13; indeterminate number of episodes of major depression; six episodes of hypomania; alcoholism; on Lithium; positive family history
GM5911		R1640 15I	0308	33	M	WH		AFF BRO	888	2	Type II bipolar illness with major depression; onset at age 10; one episode of major depression; numerous hypomanic episodes, one treated with Lithium; history of drug abuse and alcoholism
GM6021		R1640 15I	0308	40	M	WH		AFF BRO	888	3	Onset at age 32; one episode of grief reaction-minor depression; received Valium
GM5923		R1640 15I	0308	42	M	WH		AFF BRO	888	4	Onset at age 6; multiple major depressive episodes; anti-social personality; no medication
GM6055		R1640 15I	0308	37	F	WH		AFF SIS	888	5	Type I bipolar illness; onset at age 20; chronically depressed; one episode of mania with hospitalization; on Asendin, Norpramin, and Pamelor; has received Lithium
GM6054		R1640 15I	0308	64	F	WH		AFF MOTHER	888	6	Onset at age 41; two episodes of minor depression
GM5900		R1640 15I	0308	32	M	WH		PROBAND	893	1	Depressive personality; drug abuse; schizo-affective disorder (depressed type); acute manic disorder; alcoholism; positive family history
GM6039		R1640 15I	0308	31	M	WH		AFF BRO	893	2	Drug abuse; one episode of minor depression; onset at age 20
GM6048		R1640 15I	0308	30	F	WH		AFF SIS	893	3	Type I bipolar illness; onset at age 24; three major depressive episodes with hospitalization; two manic episodes with hospitalization; on Mellaril
GM6107		R1640 15I	0308	55	M	WH		AFF FATHER	893	4	Schizo-affective disorder (depressed type); manic disorder; alcoholism; 1 episode of depression treated with ECT and chemotherapy; 1 episode of mania; onset at age 52

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - 12548 and 30920											
GM6128		R1640 15I	0308	29	F	WH		AFF SIS	893	6	Unipolar illness; onset at age 20; 3 episodes of depression treated with ECT & Lithium
GM6174	26HR	R1640 15I	0308	36	M	WH		AFF BRO	893	7	Type I bipolar illness; antisocial behavior; bereavement reaction; alcoholism; onset at age 19; 10 episodes of minor depression; 1 episode of mania
GM6713		R1640 15I	0308	42	M	WH		AFF BRO	893	8	Type I bipolar illness; onset at age 42; one episode of mania; received Lithium, Cogentin, & Haldol; required hospitalization
GM7243		R1640 15I	0308	24	F	WH		PROBAND	1010	1	At risk; clinically unaffected; see GM7244 Fibroblast; mother has type II bipolar illness
GM7261		R1640 15I	0308	21	F	WH		PROBAND	1013	1	Minor depression; one episode at age 20; father has type I bipolar illness; see GM7262 Fibroblast
GM7263		R1640 15I	0308	22	M	WH		PROBAND	1014	1	Type I bipolar illness; onset at age 20; 1 episode of depression; 1 episode of mania; hospitalized twice; on Lithium; father also has type I bipolar illness; see GM7264 Fibroblast
GM7265		R1640 15I	0308	17	F	WH		SIS	1014	2	See GM7266 Fibroblast; at risk; father and brother have type I bipolar illness
GM7278		R1640 15I	0308	19	F	WH		PROBAND	1021	1	Depressive personality with minor depression; onset at age 14; mother has type I bipolar illness; mother is dizygotic twin of mother of GM7247
GM7247		R1640 15I	0308	17	F	WH		COU	1021	2	At risk; one episode of minor depression at age 15; mother has type I bipolar illness; mother is dizygotic twin of mother of proband (GM7278); see GM7248 Fibroblast
GM7259		R1640 15I	0308	24	M	WH		PROBAND	1022	1	Indeterminate number of episodes of hypomania; onset at age 9; no hospitalization; no medication; father has type II bipolar illness with major depression; mother has minor depression; see GM7260 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
*GM5927		R1640 15I	0432	50	M	WH		PROBAND	884	1	Type I bipolar illness; onset during teens; 12 hospitalizations for manic episodes & one for depression; illness has become chronic & less responsive to chemotherapy during recent years; see GM5926 Fibroblast
GM5929		R1640 15I	0432	48	F	WH		SPOUSE	884	2	Clinically unaffected; see GM5928 Fibro.
GM5931A		R1640 15I	0432	26	M	WH		SON	884	3	At risk; clinically normal; father and two sibs have type I bipolar illness; see GM5930 Fibroblast
GM5933		R1640 15I	0432	25	F	WH		AFF DAU	884	4	Type I bipolar illness; single episode at age 24; father & sib have type I bipolar illness; see GM5932 Fibroblast
GM5935		R1640 15I	0432	23	M	WH		AFF SON	884	5	Type I bipolar illness; drug and alcohol abuse; multiple admissions; chronic in recent years; possible schizoaffective; onset at age 21; father & sib have type I bipolar illness; see GM5934 Fibroblast
GM5916		R1640 15I	0432	20	M	WH		SON	884	6	At risk; clinically unaffected; two sibs and father have type I bipolar illness
GM5937A		R1640 15I	0432	18	M	WH		SON	884	7	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5936 Fibroblast
GM5939		R1640 15I	0432	16	M	WH		SON	884	8	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5938 Fibroblast
GM5941		R1640 15I	0432	14	F	WH		DAU	884	9	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness; see GM5940 Fibroblast
GM5943		R1640 15I	0432	12	M	WH		SON	884	10	At risk; clinically unaffected; father and 2 sibs have type I bipolar illness
*GM5963B		R1640 15I	0432	86	M	WH		AFF FATHER	884	11	Probable type II bipolar illness; adult age of onset; semi-annual depressions lasting more than a week; see GM5962A Fibroblast
*GM5961		R1640 15I	0432	82	F	WH		MOTHER	884	12	Some evidence of mild lability but no psychiatric diagnosis confirmed; one son has type I bipolar illness; see GM5960 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			DISORDERS OF THE NERVOUS SYSTEM								
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
*GM5903		R1640 15I	0432	56	F	WH		SIS	884	13	Clinically normal; one brother, several nieces, and nephews have type I bipolar illness; see GM5904A Fibroblast
*GM5975A		R1640 15I	0432	55	F	WH		SIS	884	14	Clinically unaffected; one affected brother; one affected son (GM5977); spouse of GM5973; see GM5974 Fibroblast
GM5973		R1640 15I	0432	58	M	WH		AFF BRO-IN-L	884	15	Minor depressive disorder; onset early teens; has brief periods with some chronic minor depression lasting for months to a year; spouse of GM5975A; father of GM5977; see GM5972 Fibroblast
GM5971		R1640 15I	0432	61	F	WH		AFF IN-LAW	884	16	Minor depressive disorder; adult onset; brief intermittent depressive periods; paternal aunt of a type II bipolar individual; sister of GM5973; see GM5970 Fibroblast
GM5969		R1640 15I	0432	82	F	WH		IN-LAW	884	17	Clinically unaffected; two brothers have unipolar major depressive disorder; paternal grandmother of a type II bipolar individual; mother of GM5973; see GM5968 Fibroblast
*GM5993		R1640 15I	0432	52	M	WH		AFF BRO	884	18	Personality disorder, unspecified, mixed with hypomanic features and anger dyscontrol; since adulthood has some role impairment; 2 children & a brother have type I bipolar illness; see GM5994 Fibro
*GM5995A		R1640 15I	0432	50	F	WH		AFF SIS-IN-L	884	19	Major depressive disorder; onset at age 30; 2 children & mother have type I bipolar illness; history of hospitalization, chemotherapy, & suicidal thoughts; daughter of GM6013B & GM6015A; spouse of GM5993; see GM5996 Fibroblast
*GM6013B		R1640 15I	0432	77	M	WH		IN-LAW	884	20	Clinically unaffected; three affected children; father of GM5995A; spouse of GM6015A; see GM6014 Fibroblast
*GM6015A		R1640 15I	0432	74	F	WH		AFF IN-LAW	884	21	Type I bipolar illness; onset at age 24; recurrent episodes of depression & mania-hypomania; three affected children; mother of GM5995A; spouse of GM6013B; see GM6016 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
					DISORDERS OF THE NERVOUS SYSTEM						
					Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)						
*GM6023		R1640 15I	0432	47	M	WH		IN-LAW	884	22	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6013B & GM6015A; brother of GM5995A; see GM6024 Fibroblast
*GM6025A		R1640 15I	0432	46	M	WH		AFF IN-LAW	884	23	Probable unipolar illness; onset at age 30 years; one major depressive episode characterized as situational; no psychosis; mother has type I bipolar illness; son of GM6013B & GM6015A; brother of GM5995A; see GM6026 Fibroblast
*GM6027		R1640 15I	0432	41	M	WH		AFF IN-LAW	884	24	Type I bipolar illness; onset at age 27; intermittent periods of irritability and hypomania since single manic hospitalization; mother has type I bipolar illness; son of GM6013B and GM6015A; s(b of GM5995A; see GM6028 Fibroblast
*GM6017		R1640 15I	0432	39	M	WH		IN-LAW	884	25	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6013B & GM6015A; brother of GM5995A; see GM6018 Fibroblast
*GM6029B		R1640 15I	0432	37	F	WH		IN-LAW	884	26	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; daughter of GM6013B & GM6015A; sister of GM5995A; see GM6030 Fibroblast
*GM6011		R1640 15I	0432	33	M	WH		IN-LAW	884	27	Clinically unaffected; 3 affected sibs; mother has type I bipolar illness; son of GM6013B & GM6015A; brother of GM5995A; see GM6012 Fibroblast
*GM5896		R1640 15I	0432	44	M	WH		BRO	884	28	Clinically normal; brother has type I bipolar illness; father of GM5888, GM5890, GM5892, & GM5894; husband of GM5898; see GM5897 Fibroblast
GM5898		R1640 15I	0432	45	F	WH		SIS-IN-LAW	884	29	Clinically normal spouse of GM5896; mother of GM5888, GM5890, GM5892, and GM5894; see GM5899 Fibroblast
GM5905		R1640 15I	0432	33	M	WH		NEPHEW	884	30	Clinically normal; brother has type II bipolar illness; nephew and cousin of several type I bipolars; son of GM5973 & GM5975A; see GM5906 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM5983		R1640 15I	0432	27	F	WH		NIECE	884	31	Clinically unaffected; brother has type II bipolar illness; daughter of GM5973 & GM5975A; see GM5984 Fibroblast
GM5977		R1640 15I	0432	26	M	WH		AFF NEPHEW	884	32	Type II bipolar illness; son of GM5973 & GM5975A; see GM5976 Fibroblast
GM5985		R1640 15I	0432	20	F	WH		NIECE	884	33	Clinically unaffected; brother has type II bipolar illness; daughter of GM5973 & GM5975A; see GM5986 Fibroblast
GM5979		R1640 15I	0432	17	F	WH		NIECE	884	34	At risk; clinically unaffected; brother has type II bipolar illness; daughter of GM5973 & GM5975A; see GM5978 Fibroblast
GM5981		R1640 15I	0432	15	F	WH		NIECE	884	35	At risk; clinically unaffected; brother has type II bipolar illness; daughter of GM5973 & GM5975A; see GM5980 Fibroblast
*GM5987A		R1640 15I	0432	30	F	WH		AFF NIECE	884	36	Type I bipolar illness; multiple admissions; similarly affected sister; daughter of GM5993 & GM5995A; see GM5988 Fibroblast; husband is individual V-28 (GM7876) in family #1075
*GM5901		R1640 15I	0432	28	F	WH		NIECE	884	37	Clinically normal; two sibs with type I bipolar illness; daughter of GM5993 and GM5995A; see GM5902 Fibroblast
*GM5918		R1640 15I	0432	26	F	WH		NIECE	884	38	Clinically normal; two sibs with type I bipolar illness; daughter of GM5993 and GM5995A; see GM5919 Fibroblast
*GM5989		R1640 15I	0432	25	M	WH		NEPHEW	884	39	Clinically normal; two sibs with type I bipolar illness; son of GM5993 and GM5995A; see GM5990 Fibroblast
*GM5907		R1640 15I	0432	24	M	WH		NEPHEW	884	40	Clinically normal; two sibs with type I bipolar illness; son of GM5993 and GM5995A; see GM5908 Fibroblast
*GM5914		R1640 15I	0432	23	M	WH		NEPHEW	884	41	Clinically normal; two sibs with type I bipolar illness; son of GM5993 and GM5995A; see GM5915 Fibroblast
*GM5999		R1640 15I	0432	22	F	WH		AFF NIECE	884	42	Type I bipolar illness; onset at age 16 yrs; one manic episode and several major depressive episodes, each lasting from several weeks to a month; similarly affected sib; daughter of GM5993 and GM5995A; see GM6000 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
*GM5991		R1640 15I	0432	20	F	WH		NIECE	884	43	Clinically unaffected; 2 sibs have type I bipolar illness; daughter of GM5993 & GM5995A; see GM5992 Fibroblast
*GM6001		R1640 15I	0432	19	M	WH		NEPHEW	884	44	Clinically unaffected; 2 sibs have type I bipolar illness; son of GM5993 & GM5995A; see GM6002 Fibroblast
*GM5997		R1640 15I	0432	18	M	WH		NEPHEW	884	45	Clinically unaffected; 2 sibs have type I bipolar illness; son of GM5993 & GM5995A
*GM6003		R1640 15I	0432	16	F	WH		NIECE	884	46	At risk; clinically unaffected; two sibs have type I bipolar illness; daughter of GM5993 & GM5995A; see GM6004 Fibroblast
*GM6005		R1640 15I	0432	12	F	WH		NIECE	884	47	At risk; clinically unaffected; two sibs have type I bipolar illness; daughter of GM5993 & GM5995A; see GM6006 Fibroblast
GM5894		R1640 15I	0432	22	M	WH		NEPHEW	884	48	Clinically normal with possible early signs of bipolar illness; second degree relative of several type I bipolar patients; son of GM5896 and GM5898; see GM5895 Fibroblast
GM5892		R1640 15I	0432	20	M	WH		NEPHEW	884	49	Clinically normal with brief periods of minor depression; second degree relative of several type I bipolar patients; son of GM5896 & GM5898; see GM5893 Fibro.
GM5888		R1640 15I	0432	18	M	WH		NEPHEW	884	50	Clinically normal; second degree relative of several type I bipolar patients; son of GM5896 & GM5898; see GM5889 Fibro
GM5890		R1640 15I	0432	16	M	WH		NEPHEW	884	51	Clinically normal; second degree relative of several type I bipolar patients; son of GM5896 & GM5898; see GM5891 Fibro
GM7845		R1640 15I	0432	84	M	WH		IN-LAW	884	52	Clinically normal; wife had recurrent major depressive disorder; wife was aunt of GM6015A; father of GM7848A, GM7847, & GM7846A
GM7848A		R1640 15I	0432	52	F	WH		IN-LAW	884	53	Clinically normal; deceased mother had recurrent major depressive disorder; daughter of GM7845; mother was aunt of GM6015A

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
				DISORDERS OF THE NERVOUS SYSTEM							
				Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)							
GM7847		R1640 15I	0432	49	F	WH		IN-LAW	884	54	Clinically normal; deceased mother had recurrent major depressive disorder; daughter of GM7845; mother was aunt of GM6015A
GM7846A		R1640 15I	0432	42	F	WH		IN-LAW	884	55	Clinically normal; deceased mother had recurrent major depressive disorder; daughter of GM7845; mother was aunt of GM6015A
GM7815A		R1640 15I	0432	74	F	WH		IN-LAW	884	56	Clinically normal; sister of GM6015A
GM7789A		R1640 15I	0432	72	M	WH		AFF IN-LAW	884	57	Minor depressive disorder & anxiety episodes; brother of GM6015A; father of GM7790A, 7813A, 7812A, 7814, & 7816A
GM7788		R1640 15I	0432	71	F	WH		IN-LAW	884	58	Clinically normal; wife of GM7789A; mother of GM7790A, 7813A, 7812A, 7814, & 7816A
GM7790A		R1640 15I	0432	53	F	WH		AFF IN-LAW	884	59	Atypical psychosis with early affective features; presently illness has some schizophrenic symptoms; may have 2 illnesses; onset at age 28; parents are GM7788 & GM7789A
GM7813A		R1640 15I	0432	51	M	WH		IN-LAW	884	60	Clinically normal; parents are GM7788 & GM7789A
GM7812A		R1640 15I	0432	50	F	WH		IN-LAW	884	61	Clinically normal; parents are GM7788 & GM7789A
GM7814		R1640 15I	0432	46	F	WH		IN-LAW	884	62	Clinically normal; parents are GM7788 & GM7789A
GM7816A		R1640 15I	0432	38	M	WH		IN-LAW	884	63	Clinically normal; parents are GM7788 & GM7789A
GM7840		R1640 15I	0432	65	F	WH		IN-LAW	884	64	Clinically normal; sister of GM6015A
GM7836		R1640 15I	0432	76	M	WH		AFF IN-LAW	884	65	Recurrent major depressive disorder; intermittent minor depressive disorder; onset at age 59; 2 sibs & mother (aunt of GM6015A) diagnosed with major depression; bro of GM7943A & GM7949B; daugh is individual IV-13 (GM7875) in family 1075
GM7949B		R1640 15I	0432	69	F	WH		AFF IN-LAW	884	66	Recurrent major depressive disorder; onset in 20s; 2 sibs & mother (aunt of GM6015A) diagnosed with major depression; sister of GM7836 & GM7943A

REP ID	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM7943A		R1640 15I	0432	74	F	WH		AFF IN-LAW	884	67	Intermittent minor depressive disorder; two siblings and mother diagnosed with major depression; mother was aunt of GM6015A; sister of GM7836 & GM7949B
GM7779C		R1640 15I	0432	70	F	WH		PAT COU	884	68	Clinically normal; three siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband
GM7777		R1640 15I	0432	69	M	WH		AFF PAT COU	884	69	Intermittent depressive disorder; probable major depressive disorder; chronic hypomania; probable type I bipolar; 2 sibs & father (paternal uncle of proband) have affective disorders; clinically normal mother was aunt of GM6015A
GM7778C		R1640 15I	0432	67	M	WH		AFF PAT COU	884	70	Atypical bipolar disorder; labile personality; two siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband
GM7776C		R1640 15I	0432	66	F	WH		PAT COU	884	71	Clinically normal; three siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband
GM7948A		R1640 15I	0432	63	M	WH		PAT COU	884	72	Clinically normal; three siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband
GM7837		R1640 15I	0432	60	F	WH		PAT COU	884	73	Clinically normal; three siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband
GM7811		R1640 15I	0432	59	F	WH		AFF PAT COU	884	74	Recurrent major depressive disorder; labile personality; two siblings and father diagnosed with affective disorders; clinically normal mother was aunt of GM6015A; affected father was paternal uncle of proband

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)											
GM7794		R1640 15I	0432	61	M	WH		IN-LAW	884	75	Clinically normal; father of GM7798, 7944B, 7797, 7796, 7942, & 7795
GM7798		R1640 15I	0432	34	F	WH		PAT 2ND COU	884	77	Clinically normal; daughter of GM7794
GM7944B		R1640 15I	0432	29	F	WH		PAT 2ND COU	884	78	Clinically normal; daughter of GM7794
GM7797		R1640 15I	0432	27	M	WH		PAT 2ND COU	884	79	Clinically normal; son of GM7794
GM7796		R1640 15I	0432	25	F	WH		PAT 2ND COU	884	80	Clinically normal; daughter of GM7794
GM7942		R1640 15I	0432	22	M	WH		PAT 2ND COU	884	81	Clinically normal; son of GM7794
GM7795		R1640 15I	0432	20	F	WH		AFF PAT COU	884	82	Bipolar schizoaffective disorder; probable schizophrenia; onset at age 15; daughter of GM7794
GM7835		R1640 15I	0432	79	F	WH		PROBAND	1075	1	Recurrent major depressive disorder; father, half-sister, paternal grandfather, & paternal uncle had type I bipolar illness; deceased brother had major depression; onset at age 37
GM7775		R1640 15I	0432	86	F	WH		SIS	1075	2	Clinically normal; wife of GM7774; deceased half-sister & father had type I bipolar illness; deceased brother had major depression; daughter (GM7785) has a manic disorder
GM7774		R1640 15I	0432	82	M	WH		IN-LAW	1075	3	Clinically normal; one deceased brother had bipolar illness; another brother had depression; a daughter (GM7785) has a manic disorder; spouse of GM7775
GM7849		R1640 15I	0432	68	F	WH		HALF-SIS	1075	4	Clinically normal; same father as proband; a daughter has type II bipolar illness; a sister has type I bipolar illness
GM7785		R1640 15I	0432	50	F	WH		AFF NIECE	1075	5	Manic disorder; psychotic subtype; onset at age 29; daughter of GM7775 & GM7774; mother of GM7783, 7787, & 7786
GM7784		R1640 15I	0432	51	M	WH		IN-LAW	1075	6	Clinically unaffected; spouse of GM7785; father of GM7783, 7787, & 7786
GM7874		R1640 15I	0432	56	M	WH		AFF SON	1075	7	Intermittent minor depressive disorder; spouse of GM7875; father of GM7876

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAM#2	REMARKS
DISORDERS OF THE NERVOUS SYSTEM												
Primary Affective Disorder - Old Order Amish (See Appendix for Pedigree)												
GM7875		R1640 15I	0432	53	F	WH			DAU-IN-LAW	1075	8	Clinically unaffected; spouse of GM7874; family history of affective disorder; father has recurrent major depression; father is individual II-33 (GM7836) in family 884; mother of GM7876
GM7783		R1640 15I	0432	25	F	WH			AFF MAT COU	1075	9	Major depressive disorder; probably psychotic & hypomanic; probably type II bipolar illness; onset at age 24; daughter of GM7784 & GM7785
GM7787		R1640 15I	0432	19	F	WH			MAT GT NIECE	1075	10	At risk; mother has manic disorder; sister has probable type II bipolar illness; daughter of GM7784 & GM7785
GM7786		R1640 15I	0432	15	M	WH			MAT COU	1075	11	At risk; mother has manic disorder; sister has probable type II bipolar illness; son of GM7784 & GM7785
GM7876		R1640 15I	0432	32	M	WH			GRANDSON	1075	12	Clinically unaffected; son of GM7874 and GM7875; paternal & maternal grandparents have major depressive disorder; wife is individual IV-17 (GM5987A) in family 884
Schizophrenia - 18150												
GM1793	20HR	R1640 20I	0137	26	M	WH	+-		PROBAND	119	2	See GM1792 Fibroblast; HLA type Aw24,B18,B13; episodes of agitation, delusions of persecution, fear of assassination; father and sister also affected
GM1836	38HR	R1640 20I	0137	27	F	WH	+-		AFF SIS	119	3	See GM1835 Fibroblast; HLA type A28,Bw16/A3,Bw35; hospitalized; drug abuse; father also affected; schizo-affective disorder
*GM1488	19HR	R1640 20I	0137	23	M	WH	+-		PROBAND	176	1	HLA type Aw26,B16/Aw26,B16; see GM2497 Fibroblast; paralogical thinking, affective shielding, splitting of affect from content, suspiciousness; onset at age 15; hospitalized; Neutral Alpha-glucosidase C phenotype=1; pos. family hx.
*GM1487	20HR	R1640 20I	0137	53	F	WH	++		MOTHER	176	2	Unaffected; HLA type Aw26,B16/A2,B35; described as "unreasonable and demanding" by family members; Neutral Alpha-glucosidase C phenotype=1
*GM1489	20HR	R1640 20I	0137	55	M	WH	+-		AFF FATHER	176	3	HLA type Aw26,B16/A18,B-; 3 episodes of depression; ECT; no hypomania; Neutral Alpha-glucosidase C phenotype=2-1; two affected children

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF THE NERVOUS SYSTEM											
Schizophrenia - 18150											
GM1490	30HR	R1640 20I	0137	27	F	WH	+-	AFF SIS	176	4	HLA type Aw26,316/A2,B35; see GM2503 Fibroblast; anorexia nervosa since adolescence; more schizoid than depressed; affected father and brother
OPHTHALMOLOGIC DISORDERS											
Bardet-Biedl Syndrome - 20990											
GM5952		R1640 15I	0433	18	M	WH	--	PROBAND	891	1	Retinal degeneration; extinguished electro-retinograms; polydactyly; partial syndactyly; obesity at 5.5 years; similarly affected brother; see GM5948 Fibro
GM5951		R1640 15I	0433	50	F	WH	+-(O)	MOTHER	891	2	Clinically unaffected; 2 affected sons; see GM5949 Fibroblast
GM5953		R1640 15I	0433	19	M	WH	--	PROBAND	892	1	Retinal degeneration; extinguished electro-retinogram; obesity; polydactyly; mental retardation; see GM5950 Fibro.
GM5954		R1640 15I	0433	50	F	WH	+-(O)	MOTHER	892	2	Clinically unaffected
GM6080		R1640 15I	0433	26	F	WH	--	PROBAND	905	1	Retinal degeneration, medullary sponge kidney, & slight mental retardation; an affected sister is in addition obese & manifests polydactyly
GM6082		R1640 15I	0433	46	F	WH	+-(O)	MOTHER	905	2	Clinically unaffected; 2 affected daughters; see GM6081 Fibroblast
GM6195		R1640 15I	0433	34	M	WH	--	PROBAND	925	1	Pigmentary retinal degeneration; abnormal electro-retinograms; bilateral macular degeneration; posterior subcapsular lens opacities; mental retardation; nystagmus; polydactyly; see GM6196 Fibro.
GM6197		R1640 15I	0433	66	F	WH	+-(O)	MOTHER	925	2	Clinically unaffected
Total Colorblindness (Achromatopsia; Day Blindness; Nyctalopia) - 21690											
GM7087		R1640 15I	0092	23	M		--	PROBAND	992	1	Photophobia; reduced vision; nystagmus; normal fundus; parents are 1st cousins; 46,XY
Foveal Dystrophy, Progressive - 13655											
GM3784		R1640 20I	0351	49	M	WH	+-	PROBAND	534	1	20/70 vision, photophobia; positive family history; Deutman variant; relative scotomas in both eyes; see GM3785 Fibro.

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OPHTHALMOLOGIC DISORDERS											
Leber Optic Atrophy - 30890											
GM3857		R1640 20I	0351	33	M	WH	Y-	PROBAND	567	1	Onset in mid-teens; decreased bilateral visual acuity; similarly affected brother; see GM3858 Fibroblast
Amaurosis Congenita of Leber; Type I and II - 20400 & 20410											
GM3851		R1640 20I	0351	29MO	F	WH	--	PROBAND	564	1	Poor vision, nystagmus, 7 diopters hyperopic; similarly affected half sister but fathers were brothers; see GM3852 Fibroblast
Leopard Syndrome (Cardiomyopathic Lentiginosis) - 15110											
GM4106		R1640 15I	0351	14	M	WH	+-	PROBAND	646	1	Part of original family described by Gorlin; see GM4107 Fibroblast; mother similarly affected; growth retardation; sensory neural hearing loss; mild pulmonary stenosis; abnormal EKG
Macular Degeneration, Juvenile (Stargardt Disease) - 24020											
GM5365		R1640 15I	0351	14	M	WH	--	PROBAND			Loss of visual acuity since age 11; consanguineous parents; normal photopic and scotopic ERG; pigmentary retinopathy; also has Kartagener syndrome (bronchiectasis and situs inversus totalis)
Ophthalmoplegia, Pigment, Degen. of Retina & Cardiomyopathy - 16510											
GM6224	25HR	R1640 15I	0446	10	M	WH	+-	PROBAND	931	1	Kearns-Sayre syndrome; external ophthalmoplegia with retinal degeneration, ptosis, myocardial fibrosis with congestive heart failure, muscle weakness, diabetes, and absence of sweat; parents have normal ERGs & EKGs; see GM6225 Fibro.
Pterygium of Conjunctiva and Cornea - 17800											
GM4181		R1640 15I	0351	34	M	WH	+-	PROBAND	681	1	Unilateral; 4 affected generations; onset at age 12; see GM4180 Fibroblast
Retinoblastoma, Hereditary - 18020											
GM3641		R1640 20I	0320	4	F	WH	+-	PROBAND	431	1	Bilateral; also pinealoblastoma; mother also has bilateral Retinoblastoma
GM3640		R1640 20I	0320	37	F	WH	+-	AFF MOTHER	431	2	Bilateral; daughter also has bilateral Retinoblastoma
GM6417		R1640 15I	0092	30	M	WH	+-	PROBAND	960	1	Persian; bilateral, familial Retinoblastoma; see GM6418 Fibroblast; 46,XY; 4% of cells show random chromosome loss; similarly affected son

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OPHTHALMOLOGIC DISORDERS												
Retinoblastoma; Sporadic												
GM1484		R1640 20I	0094	27MO	F	WH	+?	A	PROBAND	187	1	Bilateral; 46,XX,del(13)(pter>q14.1::q22.1>qter) in fibroblasts; see GM1142A Fibroblast and GM3022 SV40 Transformed Fibroblast
*GM5750		R1640 15I	0294	7	F	WH		A	SIB	863	1	Mental retard; no Retinoblastoma; 46,XX,rec(12),dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter)mat; unbal; trisomic for 13q12.3>13q22; nor RBC values for LDH, TPI, & PepB; normal elec phor patterns for LDH, PepB, TPI, & ESD
*GM5751		R1640 15I	0294	29	F	WH		A	PARENT	863	2	Phenotypically normal; 46,XX,dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter;13pter>13q12.3::13q22>13qter); balanced; a son had bilateral Retinoblastoma and multiple anomalies; proband not in Repository
GM7312		R1640 15I	0094	4	F		+?	A	PROBAND	1025	1	Bilateral; sporadic; negative family history; normal growth & development except for delayed speech; 46,XX,del(13)(pter>q14.1::q21.2>qter) in lymphocytes & fibroblasts; see GM5877 Fibroblast
Retinitis Pigmentosa - 18010, 26800, 31260												
GM3823A		R1640 15I	0351	50	M	WH			PROBAND	554	1	Mild optic atrophy with severe pigmentary retinopathy; ring scotoma; extinguished ERG; pedigree shows dominant inheritance; see GM3824 Fibroblast
GM3825		R1640 20I	0351	22	M	WH			AFF SON	554	2	Mild optic atrophy with moderate pigmentary retinopathy; pedigree shows dominant inheritance; see GM3826 Fibroblast
GM3832		R1640 20I	0351	56	F	WH			PROBAND	558	1	Fundus shows pigmentary degenerative type changes; nonrecordable ERG; ring scotomas both eyes; dominant inheritance; see GM3831 Fibroblast
GM3836		R1640 20I	0351	32	F	WH			AFF DAU	558	2	Mild abnormal pigmentation; subnormal ERG; dominant inheritance; see GM3835 Fibroblast
GM3834		R1640 20I	0351	32	F	WH			PROBAND	559	1	Night blindness; subnormal ERG; pedigree shows dominant inheritance; see GM3833 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			OPHTHALMOLOGIC DISORDERS								
Retinitis Pigmentosa - 18010, 26800, 31260											
GM3838		R1640 20I	0351	12	F	WH		AFF DAU	559	2	Poor night vision; moderately subnormal ERG; dominant inheritance; see GM3837 Fibroblast
GM3894		R1640 20I	0351	36	M	WH		PROBAND	573	1	Similarly affected father and brother; probably dominant inheritance; see GM3893 Fibroblast
GM3907		R1640 15I	0351	5	M	WH		PROBAND	578	1	General tapetoretinal degeneration; pedigree suggests X-linked inheritance; see GM3906 Fibroblast
GM3911		R1640 20I	0351	41	F	WH		AFF MOTHER	578	2	High myopia; constricted visual field; pedigree suggests X-linked inheritance; extinguished ERG; see GM3910 Fibroblast
GM7235		R1640 15I	0466	34	F	WH		PROBAND	775	1	Pedigree suggests recessive inheritance; see GM7236 Fibroblast; onset at age 3; severely myopic; pigmentary degeneration; loss of visual acuity; typical fundus findings; constricted visual fields; similarly affected brother
GM7231		R1640 15I	0466	74	M	WH		FATHER	775	2	See GM7232 Fibroblast; clinically unaffected; two affected children; pedigree suggests recessive inheritance
GM7233		R1640 15I	0466	68	F	WH		MOTHER	775	3	See GM7234 Fibroblast; clinically unaffected; two affected children; pedigree suggests recessive inheritance
GM7368		R1640 15I	0466	45	M	WH		BRO	775	4	See GM7369 Fibroblast; clinically unaffected; two affected sibs; pedigree suggests recessive inheritance
GM7482		R1640 15I	0466	47	M	WH		AFF BRO	775	5	Bilateral; onset at age 5; loss of visual acuity; constricted visual fields; similarly affected sister; pedigree suggests recessive inheritance; see GM7483 Fibroblast
Usher Syndrome - Retinitis Pigmentosa + Congenital Deafness - 27690											
GM3853		R1640 20I	0351	47	M	WH	--	PROBAND	565	1	German; mildly affected; slow progression; brother is similarly affected; see GM3854 Fibroblast
GM3890A		R1640 15I	0351	24	M	WH	--	PROBAND	572	1	Progressive tapetoretinal degeneration; similarly affected sib; see GM3889 Fibroblast

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OPHTHALMOLOGIC DISORDERS												
Usher Syndrome - Retinitis Pigmentosa + Congenital Deafness - 27690												
GM3892A		R1640 20I	0351	16	M	WH	--		AFF BRO	572	2	Progressive tapetoretinal degeneration; see GM3891 Fibroblast
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Bombay Phenotype - 21110												
GM2979	18HR	R1640 20I	0058		F	WH	--		PROBAND			Clinically normal; presence of anti-H in plasma
Esterase D Deficiency - 13328												
GM7890		R1640 15I	0312	3	F	WH		A	PROBAND			Developmental delay; growth retardation; mild facial dysmorphism; normal eye exam; 50% of normal Esterase D activity in blood; 46,XX,del(13)(pter>q12.3::q21.2>qter)
Fragile Site 10q25 - 13662												
*GM5523A		R1640 15I	0418	28	F	WH		A	PROBAND	855	1	46,XX,fra(10)(q25) in 16% of cells with FudR; see GM5425A Fibroblast; short stature, late menarche, irregular menses, small uterus, small ovaries, aortic insufficiency; myopia, and cataracts
Fumarate Hydratase, Soluble & Mitochondrial, FH-1 & FH-2 - 13685 & 13686												
GM7158		R1640 15I	0491	2	M	OT		A	PROBAND	876	1	Severely retarded and dysmorphic; Fumarate hydratase activity consistent with trisomy for 1q; 46,XY,-2,+der(2),t(1;2)(2pter>2q37::1q32.1>1qter)mat; unbalanced
Chromosomes with HSRs												
*GM4761A		R1640 15I	0310	58	M	WH		A	PROBAND			Clinically normal; 46,XY,14p+; 10% of cells show random chromosome loss; p+ appears to be a homogeneously staining region; HSR contains amplified genes for ribosomal RNA
Porphyria: Hereditary Coproporphyria - 12130												
GM2302	21HR	R1640 20I	0112	59	F	WH	+-		PROBAND	641	1	Clinically affected; see GM0962 Fibroblast
Porphyria: Acute Intermittent Porphyria - 17600												
GM2124	22HR	R1640 20I	0112	32	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; active AIP
GM2213	30HR	R1640 20I	0112	62	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; active AIP

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
OTHER DISORDERS OF KNOWN BIOCHEMISTRY												
Porphyria: Acute Intermittent Porphyria - 17600												
GM2229A		R1640 15I	0112	23	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; latent carrier
GM2230	32HR	R1640 20I	0112	58	F	WH	+-	A	PROBAND			Deficient uroporphyrinogen-I synthetase; latent carrier
*GM2135	22HR	R1640 20I	0112	34	F	WH	+-	A	PROBAND	82	1	See GM0939 Fibroblast; deficient uroporphyrinogen-I synthetase; active AIP; Neutral Alpha-glucosidase C phenotype=1; positive family history
*GM2134	24HR	R1640 20I	0112	63	F	WH	++	A	MOTHER	82	2	See GM0940 Fibroblast; clinically normal; no URO synthetase deficiency; Neutral Alpha-glucosidase C phenotype=3-1
*GM2133	45HR	R1640 20I	0112	65	M	WH	+-	A	FATHER	82	3	See GM0941 Fibroblast; deficient URO synthetase; latent carrier; Neutral Alpha-glucosidase C phenotype=1
GM1363	28HR	R1640 20I	0112		F	WH	+-	A	PROBAND	912	1	Deficient uroporphyrinogen-I synthetase; active AIP; similarly affected sister
Porphyria: Variegata - 17620												
GM2303	19HR	R1640 20I	0112	59	F	WH	+-		PROBAND			Clinically affected
Protoporphyria; Erythropoietic; Heme Synthetase Deficiency - 17700												
*GM6221		R1640 15I	0112	13	F	WH	+-		PROBAND			Mild photosensitivity and markedly elevated protoporphyrin IX in red cells and feces; PHA/PWM stimulated lymphocytes accumulate protoporphyrin IX following incubation with delta-aminolevulinic acid; deficient ferrochelatase activity
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Acute Lymphocytic Leukemia												
*GM2219C	27HR	R1640 15I	0260	19	M				PROBAND			Line Molt-4; T lymphoblast cell line; all cells show inv(2), inv(5), and 7p+; 10% of cells are diploid & 90% are tetraploid; culture is pos. for T cell 65,000 M.W. glycoprotein & neg. for sIg & Ia (B cell) antigen
*GM3671B	30HR	R1640 15I	0260	3	F	WH			PROBAND			T cell line CCRF-CEM; virus particle free; pos. for T cell 65,000 M.W. glycoprotein & neg. for sIg & Ia (B cell) antigen; all cells show 8p-, -18, inv(X), & +20; 6% of cells are diploid, 94% tetraploid; all show random chromo. abnorm.

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Acute Lymphocytic Leukemia												
*GM3638	27HR	R1640	20I 0341	16	F				PROBAND	195	1	B cell line 8392; same pt. as GM3639A; pos. for sIg & neg. for T cell 65,000 M.W. glycoprotein; all cells show 8p- & 12 p+; 2% are diploid, 90% tetraploid, & 8% polyploid with random chromosome abnormalities
*GM3639A	32HR	R1640	15I 0341	16	F				PROBAND	195	1	T cell line 8402 ; same pt. as GM3638; pos. for T cell 65,000 M.W. glycoprotein & neg. for sIg & Ia (B cell) antigen; all cells show 6q- & 11p+; 18% are near diploid, 72% are hypotetraploid, & all show random chromosome abnormalities
*GM4154A	26HR	R1640	20U 0341	11	M	WH			PROBAND	671	1	B cell line CCRF-SB; same patient as GM4155; negative for T cell 65,000 M.W. glycoprotein & positive for sIg & Ia (B cell) antigen; all cells nearly tetraploid with random chromosome loss/gain; no abnormal chromosomes
*GM4155	28HR	R1640	20U 0341	11	M	WH			PROBAND	671	1	T cell line CCRF-HSB2; same patient as GM4154A; negative for T cell 65,000 M.W. glycoprotein, sIg, & Ia(B cell) antigen; all cells nearly diploid with frequent chromosome loss; 13q-(?) marker chromosome present in all cells
AGR Triad (Aniridia, Genitourinary Abnormalities, Mental Retardation)- 19407												
*GM6803		R1640	15I 0107	18	M	WH		A	PROBAND			Aniridia, mental retardation, hypogonadism, abnormal rt. kidney, high arched palate, microphallus, cryptorchidism, & dysplastic testes; 50% of normal mother's catalase activity; 46,XY,del(11)(pter>p13::p11.2>qter)
*GM5912A		R1640	15I 0294	20	F	WH		A	PARENT	889	1	Balanced carrier; 46,XX, inv ins(11)(pter>p14::p11.2>q22::p14>p11.2::q22>qter); a daughter had aniridia, multiple anomalies, failure to thrive, & severe psychomotor retardation; proband not in Repository
*GM5913A		R1640	15I 0294	3	M	WH		A	SIB	889	2	Macular dysfunction, cleft lip and palate, & developmental delay; has triplication of 11p11.2>p14; 157% of normal RBC catalase activity; 46,XY,rec(11)(pter>q22::p14>p11.2::q22>qter)mat; a sib had aniridia

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAMS	FAMM	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
AGR Triad (Aniridia, Genitourinary Abnormalities, Mental Retardation)- 19407											
GM7736		R1640 15I	0148	1	M	WH	A	PROBAND	1074	1	Micropenis; cryptorchidism; bilateral aniridia; optic nerve atrophy; see GM6938B Fibroblast; 46,XY,del(11)(pter>p13::p14.1>qter)
Allergic Asthmatic											
GM0604	37HR	R1640 20I	0025	10	M	WH		PROBAND			Clinically affected
Antitrypsin Deficiency of Plasma (Alpha-1-Antitrypsin) - 10740											
GM3580	28HR	R1640 20I	0190	34	M	WH	+-	FATHER	209	1	Pi type MZ in serum; Greek
GM3579	44HR	R1640 20I	0190	31	F	WH	+-	MOTHER	209	2	Pi type MZ in serum; Cuban
GM3578	28HR	R1640 20I	0190	3	F	WH	--	PROBAND	209	3	Pi type ZZ in serum; Cuban-Greek
Basal Cell Nevus Syndrome (Odontogenic Keratocysts & Skeletal Anom.) - 10940											
GM1656	29HR	R1640 20I	0104	53	M	WH	+-	PROBAND	88	1	See GM1657 & GM1658 Fibroblast; clinically affected
GM1726	23HR	R1640 20I	0104	58	F	WH	+-	PROBAND	89	1	See GM1725 Fibroblast; HLA type A28,Aw30,B5,Bw16; clinically affected
GM2099	15HR	R1640 20I	0104	31	M	WH	+-	PROBAND	90	1	See GM2098 Fibroblast; clinically affected
GM2139	20HR	R1640 20I	0104		F	WH	+-	PROBAND	91	1	See GM2138 Fibroblast; clinically affected
GM1578	34HR	R1640 20I	0104	39	M	WH	+-	PROBAND	372	1	See GM1577A Fibroblast; clinically affected
GM1576	29HR	R1640 20I	0104	31	F	WH	+-	PROBAND	373	1	See GM1575 Fibroblast; clinically affected
GM1553	24HR	R1640 20I	0104	27	M	WH	+-	PROBAND	637	1	See GM1552 Fibroblast; HLA type A28,A3,Bw22,B13(B12); clinically affected
Burkitt Lymphoma											
*GM4671	24HR	R1640 10U	0051	11	M	BL		PROBAND			RAJI; ATCC CCL 86; G6PD type B; negative for EB virus; negative for reverse transcriptase; 46,XY, 4q+, 9p+, 14q+; 46% of cells show random chromosome loss or gain
*GM4678	20HR	R1640 15I	0051		M	BL		PROBAND			African; JIYOYE (P-2003); G6PD type B; pos. for EB virus; neg. for reverse transcriptase; 46,XY,mar(14),i(9)(pter>cen>pter),-14,+der(14),t(9;14)(14pter>14q32::9q11>9qter); HSR in der(14)

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Burkitt Lymphoma											
*GM4679	19HR	R1640 10U	0051	3	M	BL		PROBAND			Line EB-3; G6PD type A; positive for EB virus; negative for reverse transcriptase; 46+XY+mar(8)+mar(14); 10% of cells show random chromosome loss; band 8q23 is absent; 14q+ is unidentified
Carbonic Anhydrase, Erythrocyte; Electrophoretic Variant - 11480											
*GM2744	20HR	R1640 20I	0234	17	F	WH	--	PROBAND	629	1	Presumed homozygous for red cell carbonic anhydrase I deficiency; clinically normal; Greek
*GM2742	21HR	R1640 20I	0234	44	M	WH	+-(O)	FATHER	629	2	Greek; clinically normal
*GM2743	17HR	R1640 20I	0234	37	F	WH	+-(O)	MOTHER	629	3	Greek; clinically normal
*GM2745	22HR	R1640 20I	0234	16	M	WH	--	AFF BRO	629	4	Presumed homozygous for red cell carbonic anhydrase I deficiency; clinically normal; Greek
Chediak-Higashi Syndrome (Natural Killer Lymphocyte Defect) - 21450											
*GM3365	20HR	R1640 20I	0227	27	M	WH	--	PROBAND			Albinism; recurrent skin infections; neuropathy; mild mental retardation
GM2431A		R1640 20I	0216	32	M	WH	--	PROBAND	630	1	Nystagmus & peripheral neuropathy; formerly GM2378
GM2430	37HR	R1640 20I	0216	55	F	WH	+-(O)	MOTHER	630	2	Italian; clinically unaffected
GM2432	34HR	R1640 20I	0216	28	F	WH	+?	SIS	630	3	Clinically normal; possible heterozygote
Chronic Myelogenous Leukemia											
*GM4680	19HR	R1640 10U	0051		F			PROBAND			Line IM9; G6PD type B; possesses surface receptors for human growth hormone, insulin, and calcitonin; synthesizes IgG; negative for reverse transcriptase; 60% 46,XX/40% 92,XXXX with random chromosome loss/gain
Coffin-Lowry Syndrome - 30360											
GM3317	26HR	R1640 20I	0107	8	M	WH	Y-	PROBAND	510	1	See GM3318 Fibroblast; mental retardation; coarse facies; positive family history
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM0504	42HR	R1640 20I	0025	19	M	WH	--	PROBAND			Cell free supernatant produces ciliary dyskinesis factor in rabbit bioassay; metachromasia positive

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM0505B		R1640 20I	0025	37	F	WH	+-(O)	PARENT			Cell free supernatant produces ciliary dyskinesis factor in rabbit bioassay; metachromasia positive; proband not in Repository
GM1444	31HR	R1640 20I	0008	32	F		+-(O)	PARENT			Culture produces ciliary dyskinesis factor; proband not in Repository
GM1530	30HR	R1640 20I	0008	5	M		--	PROBAND			HLA type A2,A11,Bw17,Bw35; metachromasia positive
GM1445	37HR	R1640 20I	0008	3	F	WH	--	PROBAND	175	1	Culture produces ciliary dyskinesis factor; metachromasia negative; one similarly affected sib; Jewish descent
*GM1443	33HR	R1640 20I	0008	28	F	WH	+-(O)	MOTHER	175	2	Culture produces ciliary dsykinesis factor; metachromasia negative; Polish-Russian descent; two affected children
GM1442A		R1640 15I	0008	31	M	WH	+-(O)	FATHER	175	3	Culture produces ciliary dyskinesis factor; metachromasia negative; Hungarian-Polish-Russian descent; two affected children
GM0897	24HR	R1640 20I	0089	13	M	WH	--	PROBAND	404	1	Mild; no pancreatic insufficiency; first diagnosed at age 4 mos; a sib died from C.F. at age 10; see GM0768 Fibroblast
*GM1531	20HR	R1640 20I	0008	23	M	WH	--	PROBAND	426	1	HLA type A2,(A9),B7,(Bw40); see GM0851A Fibroblast; clinically affected
GM4330	15HR	R1640 15I	0283	20	M	BL	--	PROBAND	711	1	Classic C.F. with pancreatic insufficiency; elevated sweat electrolytes; see GM4331 Fibroblast
GM4345	22HR	R1640 15I	0283	25	M	WH	--	PROBAND	712	1	Classic C.F. with pancreatic insufficiency & diabetes mellitus; elevated sweat electrolytes; see GM4346 Fibro.
GM4540		R1640 15I	0060	10	M	WH	--	PROBAND	765	2	Abnormal sweat electrolytes; minimal cough and congestion; mild pulmonary symptomatology; little or no stool proteolytic enzyme activity; similarly affected sib; see GM4541 Fibroblast
GM7228		R1640 15I	0006		M		--	PROBAND	774	1	Clinically affected; two affected sibs
GM7227		R1640 15I	0006	6	M		--	AFF BRO	774	2	Two affected sibs; positive sweat test; bronchitis & pneumonia; frequent antibiotic treatment for pseudomonas; on catazyme therapy

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM7226		R1640 15I	0006	2	F		--	AFF SIS	774	3	Two affected sibs; positive sweat test; on catazyme therapy
GM7224		R1640 15I	0006		F		+-(0)	MOTHER	774	4	Clinically normal; 3 affected children
GM7225		R1640 15I	0006		M		+-(0)	FATHER	774	5	Clinically normal; 3 affected children
GM7337		R1640 15I	0006	55	F	WH	+-(0)	MOTHER	1028	1	Clinically normal; 3 affected children
GM7338		R1640 15I	0006	61	M	WH	+-(0)	FATHER	1028	2	Clinically normal; 3 affected children
GM7339		R1640 15I	0006	26	F	WH	--	PROBAND	1028	3	Two affected sibs; positive sweat test; chronic respiratory infections; no true C.F. pathological findings at age 6
GM7480B		R1640 15I	0006	29	F	WH	--	AFF SIS	1028	4	Two affected sibs; positive sweat test at age 9 with no findings of severe pulmonary or gastrointestinal disease
GM7621		R1640 15I	0006	21	M	WH	--	AFF BRO	1028	5	Two affected sibs; positive sweat test at age 2; mildly progressive bilateral disease
GM7381		R1640 15I	0499	11	M	WH	--	PROBAND	1038	1	Three sibs also affected
GM7380		R1640 15I	0499	17	M	WH	+?	BRO	1038	2	Clinically unaffected; 4 affected sibs
GM7382		R1640 15I	0499	16	F	WH	--	AFF SIS	1038	3	Three sibs also affected
GM7383		R1640 15I	0499	29	M	WH	--	AFF BRO	1038	4	Three sibs also affected
GM7385		R1640 15I	0499	26	F	WH	+?	SIS	1038	5	Clinically unaffected; 4 affected sibs
GM7386		R1640 15I	0499	24	M	WH	+?	BRO	1038	6	Clinically unaffected; 4 affected sibs
GM7384		R1640 15I	0499	55	M	WH	+-(0)	FATHER	1038	7	Clinically normal; 4 affected children
GM7387		R1640 15I	0499	55	F	WH	+-(0)	MOTHER	1038	8	Clinically normal; 4 affected children
GM7526		R1640 15I	0499	27	M	WH	--	AFF BRO	1038	9	Positive sweat test; bronchiectasis; aspermia; no pancreatic disease; three affected sibs
GM7469		R1640 15I	0026	15	F	WH	--	PROBAND	1044	1	Diagnosed at age 6 on basis of abnormal stools, persistent cough & rales, weight loss, & positive sweat test; receiving Pancrease, bronchodilators, & intermittent antibiotics; 50-75%ile for weight & height; similarly affected brother

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM7458		R1640 15I	0026	11	M	WH	+?	BRO	1044	2	Clinically unaffected; possible heterozygote; two affected sibs
GM7459		R1640 15I	0026	9	M	WH	--	AFF BRO	1044	3	Diagnosed at age 2 wks when asymptomatic by positive sweat test; currently on Pancrease, bronchodilators, & intermittent antibiotics; 50%ile for height & weight; similarly affected sister
GM7460		R1640 15I	0026	40	M	WH	+-(O)	FATHER	1044	4	Clinically normal; 2 affected children
GM7461		R1640 15I	0026	37	F	WH	+-(O)	MOTHER	1044	5	Clinically normal; 2 affected children
GM7462		R1640 15I	0026	70	M	WH	+?	MAT GR FATH	1044	6	Possible heterozygote; 2 affected grandchildren
GM7464		R1640 15I	0026	35	M	WH	+?	MAT UNCLE	1044	8	Possible heterozygote
GM7465		R1640 15I	0026	34	M	WH	+?	MAT UNCLE	1044	9	Possible heterozygote
GM7466		R1640 15I	0026	39	M	WH	+?	PAT UNCLE	1044	10	Possible heterozygote
GM7467		R1640 15I	0026	36	F	WH	+?	PAT AUNT	1044	11	Possible heterozygote
GM7468		R1640 15I	0026	41	M	WH	+?	PAT UNCLE	1044	12	Possible heterozygote
GM7527		R1640 15I	0026		M	WH	+?	PAT GR FATH	1044	13	Possible heterozygote; 2 affected grandchildren
GM7441		R1640 15I	0501	20	F	BL	--	PROBAND	1047	1	Elevated sweat chloride; pulmonary involvement; two similarly affected sibs
GM7440		R1640 15I	0501	32	M	BL	--	AFF BRO	1047	2	Elevated sweat chloride; pulmonary involvement; two similarly affected sibs
GM7758A		R1640 15I	0501	29	M	BL	--	AFF BRO	1047	3	Elevated sweat chloride; expired at age 29; two similarly affected sibs
GM7555		R1640 15I	0501		F	BL	+?	SIS	1047	4	Normal sweat test; three affected sibs; possible heterozygote
GM7556		R1640 15I	0501		M	BL	+?	BRO	1047	5	Normal sweat test; three affected sibs; possible heterozygote
GM7557		R1640 15I	0501		F	BL	+?	SIS	1047	6	Normal sweat test; three affected sibs; possible heterozygote
GM7439		R1640 15I	0501		F	BL	+-(O)	MOTHER	1047	7	Clinically normal; 3 affected children

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM7552		R1640 15I	0006	16	F	WH	--	PROBAND	1064	1	Positive family history in both parent's families; positive sweat test; lung biopsy showed bronchiectasis, chronic interstitial inflammation, fibrosis, and mucous gland hyperplasia & focal dilation; a sib with CF died at age 8
GM7553		R1640 15I	0006		F	WH	+-(0)	MOTHER	1064	2	Two affected children; 1 affected niece & nephew; clinically unaffected
GM7674		R1640 15I	0006	38	M	WH	+-(0)	FATHER	1064	3	Two affected children; 1 affected cousin; clinically unaffected
GM7732A		R1640 15I	0513	31	F	WH	--	PROBAND	1073	1	Two sibs also affected; mild lung disease; mild maldigestion; elevated liver enzymes; normal polyps; positive sweat test
GM7731A		R1640 15I	0513	36	F	WH	+?	SIS	1073	2	Three affected sibs; possible heterozygote; clinically unaffected
GM7733		R1640 15I	0513	29	F	WH	--	AFF SIS	1073	3	Two sibs also affected; minimal lung disease; minimal maldigestion; normal polyps; positive sweat test
GM7734		R1640 15I	0513	63	F	WH	+-(0)	MOTHER	1073	4	Three affected children; identical twin sister of GM7852
GM7735		R1640 15I	0513	71	M	WH	+?	MAT UNCLE	1073	5	Clinically unaffected; three affected nieces; possible heterozygote
GM7852		R1640 15I	0513	63	F	WH	+?	MAT AUNT	1073	7	Identical twin sister of GM7734; mother of GM7853 & GM7850
GM7850		R1640 15I	0513	28	F	WH	+?	MAT COU	1073	8	Three affected cousins; daughter of GM7852; clinically unaffected
GM7853		R1640 15I	0513	38	M	WH	+?	MAT COU	1073	9	Three affected cousins; son of GM7852; clinically unaffected
GM7830		R1640 15I	0521	19	F	WH	--	PROBAND	1076	1	Two sibs are also affected; diagnosed at about age 6; moderate pulmonary and no gastrointestinal symptoms at the time of diagnosis; no symptoms present at birth; receives enzyme supplements
GM7831		R1640 15I	0521	18	M	WH	--	AFF BRO	1076	2	Two sibs are also affected; diagnosed at about age 3; elevated sweat chloride; moderate pulmonary and mild gastrointestinal symptoms at the time of diagnosis; no symptoms present at birth; receives enzyme supplements

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM7832		R1640 15I	0521	15	M	WH	--	AFF BRO	1076	3	Two sibs are also affected; diagnosed at about age 1; no pulmonary or gastrointestinal symptoms at the time of diagnosis; no symptoms present at birth; receives enzyme supplements
GM7828		R1640 15I	0521	43	M	WH	+-(O)	FATHER	1076	4	Clinically normal; 3 affected children
GM7829A		R1640 15I	0521	40	F	WH	+-(O)	MOTHER	1076	5	Clinically normal; 3 affected children
GM7824		R1640 15I	0521	20	F	WH	--	PROBAND	1078	1	Two sibs are also affected; diagnosed at about age 1; elevated sweat chloride; severe pulmonary and moderate gastrointestinal symptoms at the time of diagnosis; meconium ileus at birth; receives enzyme supplements
GM7825		R1640 15I	0521	21	F	WH	--	AFF SIS	1078	2	Two sibs are also affected; diagnosed shortly after birth; elevated sweat chloride; moderate pulmonary and severe gastrointestinal symptoms at the time of diagnosis; no symptoms present at birth; receives enzyme supplements
GM7826A		R1640 15I	0521	17	M	WH	--	AFF BRO	1078	3	Two sibs are also affected; diagnosed shortly after birth; elevated sweat chloride; no pulmonary and moderate gastrointestinal symptoms at the time of diagnosis; no symptoms present at birth; receives enzyme supplements
GM7827		R1640 15I	0521	14	F	WH	+?	SIS	1078	4	Three affected sibs; possible heterozygote; clinically unaffected
GM7822		R1640 15I	0521	43	M	WH	+-(O)	FATHER	1078	5	Clinically normal; 3 affected children
GM7823		R1640 15I	0521	40	F	WH	+-(O)	MOTHER	1078	6	Clinically normal; 3 affected children
GM7857		R1640 15I	0513	14	F	WH	--	PROBAND	1080	1	Two sibs also affected; consanguineous parents; mild disease; positive sweat test
GM7860A		R1640 15I	0513	10	M	WH	--	AFF BRO	1080	3	Two sibs also affected; consanguineous parents; pulmonary and gastrointestinal disease; positive sweat test
GM7856A		R1640 15I	0513	30	M	WH	+?	BRO	1080	4	Three affected sibs; possible heterozygote; clinically unaffected; consanguineous parents; normal sweat test

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Cystic Fibrosis (Mucoviscidosis) - 21970											
GM7858		R1640 15I	0513	13	M	WH	+?	BRO	1080	5	Three affected sibs; possible heterozygote; clinically unaffected; consanguineous parents; normal sweat test
GM7855		R1640 15I	0513	53	F	WH	+-(O)	MOTHER	1080	6	Normal sweat test; 3 affected children
GM7854A		R1640 15I	0513	54	M	WH	+-(O)	FATHER	1080	7	Normal sweat test; 3 affected children
Diabetes Mellitus of the Pima Indian - 22210											
GM2575	25HR	R1640 20I	0215	23	M	AI		PROBAND	230	1	Pima-Papago American Indian; onset at age 18; on Insulin; see GM2574 Fibro.
GM2566	30HR	R1640 20I	0215	28	M	AI		PROBAND	231	1	Onset at age 27 yrs; not on Insulin; see GM2565 Fibroblast
GM2417	48HR	R1640 20I	0215	42	F	AI		PROBAND	383	1	Mother & siblings are also affected; see GM2416 Fibroblast
Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585											
GM1905	22HR	R1640 20I	0034	32	F			PROBAND			MODY diabetes diagnosed at age 10; both parents and three brothers had MOD diabetes; HLA type A11,-/Bw35,-
*GM1247A		R1640 15I	0034	29	M	WH		PROBAND	95	1	MODY; onset at age 11; see GM1237 Fibroblast; produces IgG3; 6 affected and 4 unaffected sibs
*GM1246	16HR	R1640 20I	0034	46	M	WH		AFF BRO	95	2	MODY; onset at age 29; see GM1219 Fibro.
*GM1240	22HR	R1640 20I	0034	33	F	WH		AFF SIS	95	3	MODY; onset at age 16; see GM1122 Fibro.
*GM1243	20HR	R1640 20I	0034	35	M	WH		AFF BRO	95	4	MODY; onset at age 27; see GM1430 Fibro.
*GM1956	20HR	R1640 20I	0034	44	M	WH		AFF BRO	95	5	MODY; onset at age 25; see GM1955 Fibro.
*GM1498	19HR	R1640 20I	0034		F	WH		AFF SIS	95	6	MODY; onset at age 30; see GM1497 Fibroblast; mother of GM1242, 1244, 7976, & 7977
*GM1242	20HR	R1640 20I	0034	22	M	WH		AFF NEPHEW	95	7	MODY; onset at age 14; see GM1435 Fibroblast; son of GM1498
*GM1244	25HR	R1640 20I	0034	20	M	WH		AFF NEPHEW	95	8	MODY; onset at age 12; see GM1496 Fibroblast; son of GM1498
*GM1410	26HR	R1640 20I	0034	15	F	WH		AFF NIECE	95	9	See GM1409 Fibroblast; mat cousin of GM1242 & GM1244; onset at age 13; only JOD in family; on Insulin; HLA type A2, Aw24,Bw35,B13(B12)

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585											
*GM1838	22HR	R1640 20I	0034	22	F	WH		AFF COU	95	10	MODY type II; onset at age 18; pat. 1st cousin once removed of the 6 sibs; HLA type A28(A2),Aw30,B12(B13),Bw35; see GM1837 Fibroblast
*GM7973		R1640 15I	0034	54	F	WH		IN-LAW	95	14	Clinically unaffected; affected spouse, children, and grandchild
*GM7974		R1640 15I	0034	31	F	WH		AFF NIECE	95	15	Impaired glucose tolerance test at age 15; diabetic glucose tolerance test at age 17; treated with diet alone; still has abnormal glucose tolerance; mother of GM7975
*GM7975		R1640 15I	0034	13	F	WH		AFF RELATIVE	95	16	Fasting hyperglycemia at age 13; treated with Diabinese; daughter of GM7974; affected daughter of proband's niece
*GM7976		R1640 15I	0034	24	M	WH		NEPHEW	95	17	Diabetic glucose tolerance test at age 9; treated with diet alone; presently non-diagnostic glucose tolerance test; son of GM1498
*GM7977		R1640 15I	0034	26	M	WH		AFF NEPHEW	95	18	Diabetes, type II; MODY; son of GM1498; diabetic glucose tolerance test at age 13; treated with Diabinese
GM1241A	27HR	R1640 15I	0034	37	M	WH		PROBAND	291	1	Third generation of MOD; HLA type A3, A10,B13,B12; see GM1486 Fibroblast
Diabetes Mellitus: Juvenile Onset - 22210											
*GM2676	30HR	R1640 20I	0095	17	M	WH		PROBAND	236	1	Onset at age 2; on Insulin; HLA type A1, B8/A10,B18; similarly affected sib; see GM2637 Fibroblast
*GM2793A	117HR	R1640 20I	0260	10	F	WH		AFF SIS	236	2	Onset at age 3; on Insulin; HLA type A1, B8/A10,B18; see GM2638 Fibroblast
*GM2675	25HR	R1640 20I	0095	19	M	WH		BRO	236	3	See GM2639 Fibroblast; HLA type A1,B8/A10,B18; non-diabetic; two affected sibs
*GM2741B		R1640 15I	0095	41	M	WH		FATHER	236	4	Non-diabetic; HLA type A1,B8/A3,Bw35; see GM2640 Fibroblast; both parents had maturity onset diabetes; two affected children
*GM3355		R1640 20U	0260	41	F	WH		MOTHER	236	5	Non-diabetic; HLA type A10(25),B18/A3, B7; two affected children; see GM2641 Fibroblast

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
*GM2713A		R1640 15I	0095	17	M	WH		PROBAND	243	1	Onset at age 14; on Insulin; HLA type A1,B8/A9,B18; two affected and three unaffected sibs; see GM2712 Fibroblast
*GM2705	38HR	R1640 20I	0095	45	M	WH		FATHER	243	2	Non-diabetic; HLA type A1,B8/A2,Bw35; 3 affected children; see GM2704 Fibro.
*GM2707	37HR	R1640 20I	0095	45	F	WH		MOTHER	243	3	Non-diabetic; HLA type A9(24),B18/A2,B7; 3 affected children; see GM2706 Fibro.
*GM2709	23HR	R1640 20I	0095	16	M	WH		BRO	243	4	Non-diabetic; HLA type A2,B7/A2,Bw35; see GM2708 Fibroblast; 3 affected sibs
*GM2711	31HR	R1640 20I	0095	10	M	WH		BRO	243	5	Non-diabetic; HLA type A2,B7/A2,Bw35; see GM2710 Fibroblast; 3 affected sibs
*GM2728	43HR	R1640 20I	0095	25	F	WH		SIS	243	6	Non-diabetic; HLA type A1,B8/A9,B18; see GM2869 Fibroblast; three affected sibs
*GM3027	36HR	R1640 20I	0260	23	M	WH		AFF BRO	243	8	Onset at age 11; HLA type A1,B8/A9,B18; see GM2956A Fibroblast
*GM3235	24HR	R1640 20I	0260	14	M	WH		PROBAND	606	1	HLA type A3,Bw17/A9(23),B12; onset at age 6 yrs; on Insulin; see GM3358 Fibroblast; father is also affected
*GM3239		R1640 10U	0260	13	M	WH		BRO	606	2	HLA type A3,Bw17/A9(23),B12; non-diabetic; see GM3359 Fibroblast; affected father and brother
*GM3236	26HR	R1640 15U	0260	42	M	WH		AFF FATHER	606	3	HLA type A9(23),B12/A28,Bw15; onset at age 14 yrs; on Insulin; see GM3360 Fibroblast; son is also affected
*GM3237		R1640 10U	0260	16	M	WH		BRO	606	4	HLA type A3,Bw17/A28,Bw15; non-diabetic; see GM3361 Fibroblast; affected brother and father
*GM3238B		R1640 10U	0260	40	F	WH		MOTHER	606	5	HLA type A1,Bw40/A3,Bw17; non-diabetic; see GM3362 Fibroblast
*GM2762	22HR	R1640 20I	0095	13	F	WH		PROBAND	620	1	Onset at age 10; on Insulin; HLA type A2,Bw16/A1,B8; three affected sibs; see GM1872 Fibroblast
*GM2763	20HR	R1640 20I	0095	17	F	WH		AFF SIS	620	2	Onset at age 13; on Insulin; HLA type A10,Bw16/A1,B8; see GM1873 Fibroblast

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Diabetes Mellitus: Juvenile Onset - 22210											
*GM2765	20HR	R1640 20I	0095	21	M	WH		AFF BRO	620	3	Onset at age 4; on Insulin; HLA type A10,Bw16/A1,B8; see GM1909 Fibroblast; hypertension; short stature
*GM3354A	20HR	R1640 20U	0260	21	F	WH		SIS	620	4	Non-diabetic; HLA type A2,Bw16/A3,B7; see GM1910 Fibroblast; 4 affected sibs
*GM2764	31HR	R1640 20I	0095	22	M	WH		AFF BRO	620	5	Onset at age 4; on Insulin; HLA type A2,Bw16/A1,B8; see GM1911 Fibroblast
*GM2866A	87HR	R1640 20I	0095	19	M	WH		BRO	620	6	Non-diabetic; HLA type A2,Bw16/A1,B8; see GM1875 Fibroblast; 4 affected sibs
*GM2766	26HR	R1640 20I	0095	24	F	WH		SIS	620	7	HLA type A2,Bw16/A1,B8; mentally retarded; non-diabetic; see GM1874 Fibroblast; four affected sibs
*GM2812A	19HR	R1640 10U	0260	20	M	WH		BRO	620	8	Non-diabetic; HLA type A10,Bw16/A1,B8; see GM1876 Fibroblast; 4 affected sibs
*GM2714	50HR	R1640 20I	0095	42	F	WH		MOTHER	620	9	Non-diabetic; HLA type A1,B8/A3,B7; four affected children; see GM1878 Fibroblast
*GM3356A	30HR	R1640 10U	0260	53	M	WH		FATHER	620	10	Non-diabetic; HLA type A2,Bw16/A10,Bw16; 4 affected children; see GM1877 Fibro.
*GM2677	27HR	R1640 20I	0095	24	F	WH		PROBAND	621	1	Onset at age 6; HLA type A1,B8/ Aw31, Bw40; see GM2613 Fibroblast; similarly affected brother
*GM2678	21HR	R1640 20I	0095	27	M	WH		AFF BRO	621	2	Onset at age 18 mo.; on Insulin; HLA type A1,B8/A1,--; see GM2614 Fibroblast
*GM2679	28HR	R1640 20I	0095	29	M	WH		BRO	621	3	See GM2615 Fibroblast; HLA type A1,B8/ A1,--; non-diabetic
*GM2680	26HR	R1640 20I	0095	63	M	WH		FATHER	621	4	Non-diabetic; HLA type A1,B8/A2,Bw15; see GM2616 Fibroblast; 2 affected children
*GM2785A		R1640 15I	0095	60	F	WH		MOTHER	621	5	Non-diabetic; HLA type Aw31,Bw40/A1,--; see GM2617 Fibroblast; 2 affected children
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230											
*GM1795	25HR	R1640 20I	0189	13	F	WH		PROBAND	96	1	See GM1610 Fibroblast; HLA type Aw32, B12/Aw32,Bw40; on Insulin; onset at age 11; optic atrophy; two affected sibs

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DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Diabetes Mellitus and Insipidus with Optic Atrophy - 22230												
*GM1799	17HR	R1640 20I	0189	18	F	WH			AFF SIS	96	2	HLA type Aw32,B12/A9,Bw35; not on Insulin; onset at age 16; optic atrophy; 2 affected sibs
*GM1797	50HR	R1640 20I	0189	42	M	WH			FATHER	96	4	Non-diabetic; HLA type A9,Bw35/Aw32,Bw40; see GM1701 Fibroblast; Neutral Alpha-glucosidase C phenotype=1; three affected children
*GM1796	25HR	R1640 20I	0189	16	M	WH			BRO	96	5	Non-diabetic; HLA type Aw32,Bw40/Aw32,B12; three affected sibs
*GM1800	29HR	R1640 20I	0189	7	F	WH			SIS	96	6	Non-diabetic twin of GM1801; HLA type A2,Bw15/A9,Bw35; three affected sibs
*GM1801	27HR	R1640 20I	0189	7	F	WH			SIS	96	7	Non-diabetic twin of GM1800; HLA type A2,Bw15/A9,Bw35; three affected sibs
*GM1930	22HR	R1640 20U	0189	39	F	WH			MOTHER	96	8	Non-diabetic; HLA type A2,Bw15/Aw32,B12; formerly GM1802; three affected children
*GM1803	38HR	R1640 20I	0189	11	F	WH			SIS	96	9	Non-diabetic; HLA type Aw32,B12/A9,Bw35; three affected sibs
DiGeorge Syndrome - 18840												
GM5878		R1640 15I	0148	28	M	WH		A	PARENT			Proband not in Repository; affected dau. has unbalanced translocation; 46,XY, t(10;22)(10pter>10q26.3::22q11.2>22qter; 22pter>22q11.2::10q26.3>10qter); balanced; clinically normal
*GM7939		R1640 15I	0524	3	M	WH	+-		PROBAND			Brother and father are also affected; hypocalcemia, evidence of deficient cellular immunity; characteristic dysmorphism, absence of thymic shadow, & truncus arteriosus; normal karyotype
Dyskeratosis Congenita; Zinsser-Cole-Engman Syndrome - 30500												
GM1775	30HR	R1640 20I	0107	7	M	WH	Y-		PROBAND	99	1	See GM1774 Fibro.; HLA type Aw30,(Bw40), B13,B5; mat. grfath. was also affected; leukoplakia tongue; dysplastic finger & toenails; fibro show increased bleomycin induced chromo. instabil. & hypersens. to Mitomycin-C induced cell killing
*GM3193	29HR	R1640 20I	0295	15	M	WH	Y-		PROBAND	515	1	Mild anisopoikilocytosis & mild oval macrocytosis; 2 affected brothers & 3 affected uncles

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED		RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Dyskeratosis Congenita; Zinsser-Cole-Engman Syndrome - 30500												
*GM3194	20HR	R1640 20I	0295	18	M	WH	Y-		AFF BRO	515	2	Mild anisopoikilocytosis & mild oval macrocytosis
*GM3195	23HR	R1640 20I	0295	19	M	WH	Y-		AFF BRO	515	3	Mild anisopoikilocytosis & mild oval macrocytosis
*GM3650		R1640 20I	0295	47	F	WH	+-(D)		MOTHER	515	4	Clinically normal; 3 affected brothers & 3 affected sons
Hyper IgM												
GM0390A	35HR	R1640 20I	0010	3	F	WH			PROBAND			46,XX
Leukemia; Sub-acute												
GM2584	25HR	R1640 20I	0008	60	M	WH		B	PROBAND			46,XY; 52% of cells show random chromosome loss/gain; culture is negative for T cell 65,000 M.W. glycoprotein & positive for surface immunoglobulins and Ia (B cell) antigen
Melanoma; Malignant - 15560												
GM6921		R1640 15I	0226	30	M	WH	+-		PROBAND			B-K Mole syndrome; familial dysplastic nevus syndrome; similarly affected mother & uncle; 5 primary melanomas; multiple pigmented dysplastic nevi
Multiple Myeloma Patients - 25450												
*GM2132C		R1640 15I	0205	61	M			B	PROBAND			Line RPMI8226; ATCC CCL 155; IgG Lambda-type light chain secreting; G6PD=A; negative for reverse transcriptase; modal no. of 65 chromosomes; range = 40 to 78; low TdT activity; negative for EBV and EBNA
GM1311A	24HR	R1640 15I	0156	68	M	BL			PROBAND	180	1	Bone marrow aspirate; HLA type A10,A11 (A3),B12; same patient as GM1312A
*GM1312A		R1640 15I	0156	68	M	BL			PROBAND	180	1	Peripheral blood; HLA type A10,A11(A3),B12; same patient as GM1311A
*GM1500B		R1640 15I	0010		M				PROBAND	777	1	HLA type A2(A3),B12,Bw15; secretes IgG2 (Kappa); EBNA positive; negative for T cell 65,000 M.W. glycoprotein; positive for sIg and Ia (B cell) antigen; 78% 46,XY; 22% of cells show random chromosome loss; 4% of cells have a t(7;8)
▶*GM4672B		R1640 15U	0028		M				MUTANT	777	1	EMS induced mutant of GM1500; HPRT deficient; selectable with 6-thioguanine; secretes IgG2 (Kappa); has been used to make human-human hybridomas; line GM1500 6TG-A1 (Croce)

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Multiple Myeloma Patients - 25450											
GM6923		R1640 15I	0156	74	F	WH		PROBAND	894	1	Diabetes; bone pain; metastatic breast carcinoma; has received external radiation therapy; culture established using Epstein-Barr virus
Neurofibromatosis (von Recklinghausen Disease) - 16220											
GM1634	25HR	R1640 20I	0104	61	M	WH	+-	PROBAND	113	1	Atypical; see GM1633 Fibroblast
GM1641	30HR	R1640 20I	0104	19	F	BL	+-	PROBAND	114	1	Clinically affected; see GM1639 Fibro.
GM1861	30HR	R1640 20I	0104	41	M	WH	+-	PROBAND	115	1	HLA type A3,A2,B27,Bw22(Bw40); see also GM1858, GM1859, and GM1860A Fibroblast; cafe au lait spots and neurofibromas
GM3421		R1640 20I	0294	39	F	WH	+-	AFF MOTHER	212	1	Multiple cutaneous neurofibromas; positive family history
GM3420		R1640 20I	0294	17	F	WH	+-	PROBAND	212	2	Cafe au lait spots; neurofibromas; mental retardation; positive family history
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
GM2633	22HR	R1640 20I	0225	31	F	WH	+-	PROBAND	522	1	Many affected members in pedigree; 46,XX; 10% of cells show random chromosome loss; 18% of cells show chromosome breaks; centromeres are fragile on chromosomes #1, #9, & #16
GM3224	20HR	R1640 20I	0225	25	M	WH	+-	AFF SON	603	1	See GM3223 Fibroblast; 46,XY; 14% of cells show random chromosome loss; 8% of cells show chromosome aberrations
GM3315A	21HR	R1640 20I	0225	48	F	WH	+-	PROBAND	603	2	See GM3314 Fibroblast; adenocarcinoma of colon; 5 affected children, 3 affected sibs, & 2 affected cousins; 46,XX; 16% of cells show random chromosome loss; 20% polyploid; 4% of cells show chromosome breakage
GM3941		R1640 15I	0225	45	M	WH	+?	BRO	603	3	At risk; see GM3940 Fibroblast
GM3945		R1640 15I	0225	38	F	WH	+-	AFF SIS	603	4	Cysts, polyps, ileoproctostomy; see GM3944 Fibroblast; 46,XX; 14% of cells show random chromosome loss; 6% of cells show chromosome aberrations
GM3943		R1640 15I	0225	17	F	WH	+?	NIECE	603	5	At risk; daughter of GM3945; see GM3942 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530											
GM3947		R1640 15I	0225	22	F	WH	+-	AFF DAU	603	6	46,XX with 18% random chromosome loss/gain; 6% of cells show chromosome aberrations; see GM3946 Fibroblast
GM3949		R1640 15I	0225	30	F	WH	+-	AFF DAU	603	7	Cysts, polyps, and desmoid tumor; see GM3948 Fibroblast
GM3951A		R1640 15I	0225	12	F	WH	+?	DAU	603	8	At risk; see GM3950 Fibroblast; three affected sibs and mother
GM3953		R1640 15I	0225	20	F	WH	+?	NIECE	603	9	At risk; unaffected parents; father is GM3941; see GM3952 Fibroblast
GM3955		R1640 15I	0225	24	M	WH	+-	AFF SON	603	10	Polyps and cysts; see GM3954 Fibroblast
GM3957		R1640 15I	0225	27	F	WH	++	DAU-IN-LAW	603	11	Clinically normal; see GM3956 Fibroblast
*GM7500		R1640 15I	0468	23	M	WH	+-	PROBAND	967	1	Dental abnormalities, multiple polyposis, multiple osteomas, multiple fibromas, multiple epidermoid cysts, & desmoids; see GM6855 Fibroblast
*GM7501		R1640 15I	0468	20	F	WH	+-	AFF SIS	967	5	Polyps, desmoids, fibromas, epidermoid cysts, osteomas, dental abnormalities, & adenomas see GM6964 Fibroblast
*GM7499		R1640 15I	0468	53	F	WH	+-	AFF MOTHER	967	6	Polyps, desmoids, fibromas, epidermoid cysts, osteomas, dental abnormalities, & adenomas; affected father, brother, & children; see GM6965 Fibroblast
*GM7509		R1640 15I	0468	23	M	WH	++	MAT COU	967	7	Clinically unaffected; son of GM7511
*GM7510		R1640 15I	0468	22	F	WH	+-	AFF MAT COU	967	8	Multiple polyposis, osteomas, fibromas, & epidermoid cysts; dental abnormalities; daughter of GM7511
*GM7512		R1640 15I	0468	10	F	WH	+-	AFF MAT COU	967	10	Multiple osteomas, fibromas, & epidermoid cysts; daughter of GM7511
*GM7515		R1640 15I	0468	16	M	WH	++	MAT COU	967	11	Clinically unaffected; son of GM7511
*GM7516		R1640 15I	0468	17	F	WH	+-	AFF MAT COU	967	12	Multiple fibromas and epidermoid cysts; daughter of GM7511
*GM7517		R1640 15I	0468	14	F	WH	+-	AFF MAT COU	967	13	Multiple osteomas & fibromas; desmoids; daughter of GM7511

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530												
*GM7518		R1640 15I	0468		F	WH	++		IN-LAW	967	14	Clinically unaffected spouse of GM7511; mother of GM7509, 7510, 7512, 7515, 7516, & 7517
*GM7513		R1640 15I	0468	51	F	WH	++		MAT AUNT	967	15	Clinically unaffected; father and 2 sibs are affected; mother of GM7514
*GM7514		R1640 15I	0468	23	F	WH	++		MAT COU	967	16	Clinically unaffected; daughter of GM7513
Pseudohypoparathyroidism and Pseudo-pseudohypoparathyroidism - 30080												
GM4411A	20HR	R1640 15I	0277	33	F	WH	+-	A	PROBAND			Hypocalcemia; hypothyroidism; brachydactyly; obese; onset at age 14; responsive to Vitamin D; deficient N protein in erythrocytes, platelets & lymphoid culture; only affected family member
GM4425	35HR	R1640 15I	0277	48	F	WH	+-	A	PROBAND			Pseudo-pseudohypoparathyroidism; intermittent hypocalcemia; hypothyroidism; short stature; elevated serum PTH; normal urinary response to PTH; normal N protein in erythrocytes & lymphoid culture
*GM4426	30HR	R1640 15I	0277	56	M	WH	Y-	A	PROBAND			Brachydactylia; short stature; obesity; hypocalcemia; hypothyroidism; subcutaneous calcifications; elevated serum PTH; decreased urinary response to PTH; prolactin deficiency; resistance to ADH; reduced N protein in lymphoid culture
*GM4427	26HR	R1640 15I	0277	50	M	WH	Y-	A	PROBAND			Brachydactylia; short stature; hypocalcemia; hypothyroidism; prolactin deficient; elevated serum PTH; decreased urinary response to PTH; ADH resistance; reduced N protein in lymphoid culture
GM4428	24HR	R1640 15I	0277	30	M	WH	Y-	A	PROBAND			Hex./Amer.; intermittent hypocalcemia; prolactin deficiency; resistance to parathyroid hormone at bone and kidney; elevated serum PTH; decreased urinary response to PTH; no skeletal abnormalities; norm. N protein in lymph. culture
Prader-Willi Syndrome - 17627												
*GM4297		R1640 15I	0294	40	F	WH	+-		PROBAND			Mental retardation; obesity; hypogonadism; small hands and feet; 4 affected cousins; Acadian descendant of So. Louisiana; 16% of fibroblasts show random chromosome loss; 46,XX

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY											
Rothmund-Thomson Syndrome (Poikiloderma Atrophicans & Cataract) - 26840											
GM2451A		R1640 20I	0155	25	M	WH	--	PROBAND			Clinically affected: 46,XY; 10% of cells show random chromosome loss
Sideroblastic Anemia											
GM4515	26HR	R1640 15I	0074	5	F	WH		PROBAND	736	1	Pancytopenia, pancreatic insufficiency, dystonia, and neurological symptoms; see GM4516 Fibroblast
Smith-Lemli-Opitz Syndrome (RSH Syndrome) - 27040											
GM5789		R1640 15I	0204	17MO	F	WH	--	PROBAND			46,XX in leukocytes; failure to thrive, developmental delay, hypotonia, microcephaly, micrognathia, simian creases, & partial syndactyly; negative family history; see GM5788 Fibroblast
Tuberous Sclerosis - 19110											
GM3549		R1640 20I	0006	16	M	WH	+-	PROBAND			Positive family history; clinically affected
GM1636	23HR	R1640 20I	0104	17	M	BL	+-	PROBAND	124	1	See GM1643 and GM1635 Fibroblast; clinically affected
GM1638	23HR	R1640 20I	0104	20	F	BL	+-	PROBAND	125	1	See GM1644 and GM1637 Fibroblast; clinically affected
GM7421		R1640 15I	0500	3	F	WH	+-	PROBAND	1036	1	Seizures since age 2; white spots; shagreen patch; CT scan showed brain calcifications; receives Tegretol
GM7416		R1640 15I	0500	30	F	WH	++	MOTHER	1036	2	Clinically unaffected
GM7415		R1640 15I	0500	37	M	WH	+-	AFF FATHER	1036	3	Seizures since age 4; white spots; periungual fibromas of toes; pits in tooth enamel; receives Dilantin; 5 affected sibs, affected father & daughter
GM7417		R1640 15I	0500	35	F	WH	+-	AFF PAT AUNT	1036	4	White spots; fibromas around fingernails; seizures during puberty; 5 affected sibs & father
GM7418		R1640 15I	0500	25	F	WH	+-	AFF PAT AUNT	1036	5	Seizures since puberty; white spot; shagreen patch; periungual fibroma of finger; 5 affected sibs & father
GM7419		R1640 15I	0500	29	M	WH	+-	AFF PAT UNC	1036	6	White spot; treated for seizures as a child; shagreen patches; normal fundi; 5 affected sibs & father

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY												
Tuberous Sclerosis - 19110												
GM7420		R1640 15I	0500	25	M	WH	+-		AFF PAT UNC	1036	7	White spots; periungual fibroma of finger; facial angiofibroma; no retinal lesions; shagreen patch; seizures; 5 affected sibs & father; receives Tegretol
GM7399		R1640 15I	0500	41	F	WH	++		PAT AUNT	1036	8	No white spots; no seizures; normal head CT & kidney ultrasound exams; clinically unaffected
GM7402		R1640 15I	0500	44	M	WH	++		IN-LAW	1036	9	Clinically unaffected; husband of GM7399
GM7400		R1640 15I	0500	15	F	WH	++		PAT COU	1036	10	No seizures or lesions; 2 small questionable white spots; no retinal lesions; head CT and kidney ultrasound scans were negative; daughter of GM7402 & GM7399
GM7401		R1640 15I	0500	18	F	WH	++		PAT COU	1036	11	No seizures, white spots, or lesions; head CT scan and kidney ultrasound exams were normal; daughter of GM7402 & GM7399
GM7414		R1640 15I	0500	66	M	WH	+-		AFF GR FATH	1036	12	Periungual fibromas of toes; no shagreen patches or facial angiofibroma; white spots; calcified retinal hamartoma; father of GM7415
GM7422		R1640 15I	0500	66	F	WH	++		GR MOTHER	1036	13	Clinically unaffected; wife of GM7414
Waldenstrom Macroglobulinemia - 15360												
GM1501	20HR	R1640 20I	0010	69	M		+-		PROBAND			HLA type A3,A9,Bw22,(B12); clinically affected
Wilms Tumor (Nephroblastoma) - 19407												
GM7427		R1640 15I	0094	13MO	M		+-		PROBAND			Bilateral Wilms tumor; history of scrotal hypospadias, chordee, & hydrocoele; neoplasm showed extensive mesenchymal differentiation including sheets of skeletal & smooth muscle, an island of cartilage, & islands of osteoid; 46,XY
GM3485		R1640 20I	0195	13	F	WH	+-		PROBAND			Five additional family members have Wilms tumor; 46,XX; both chromosomes #11 appear normal
*GM5518		R1640 15I	0294	31MO	F	WH	+-	A	PROBAND			Aniridia-Wilms tumor; ment retardation; failure to thrive; sporadic; no fam hist for aniridia-Wilms; 50% of RBC catalase act; nor RBC LDH-A act; nor fibro Hb epsilon DNA genes; 46,XX-9,+der(9)(9qter>9p22::11p13>11pter),del(11)(qter>p11.1:)

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS												
Chronic Granulomatous Disease (Xk-Related; CGD) - 30640 & 31485												
*GM7773		R1640 15I	0039	28	F	WH	+-	A	PROBAND			Mild ment retard; norm gonad develop; pt has partial OTC deficiency & heterozygosity for chronic granulomatous disease; lymphocytes & fibro show random X inactiv; in lymph cult, del(X) is late repl; 46,X,del(X)(pter>21.3::21.1>qter)
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955												
*GM3200	24HR	R1640 20I	0061	34	M	WH	Y-		PROBAND			46,fra(X)(q27),Y; 17% in peripheral blood leukocytes; 46,XY in lymphoid culture
*GM4025B		R1640 15I	0061	29	M	BL	Y-		PROBAND	588	1	Macro-orchidism; mental retardation; 52% 46,fra(X)(q27),Y in leukocytes; 4% in fibroblasts; fra(X) not observed in lymphoid culture; G6PD = A in blood; see GM4024A Fibroblast
GM5854		R1640 15I	0148	60	F	WH	+-(O)	A	GRMOTHER	819	4	One affected son; two affected grandsons; 1% of lymphocytes and 4% of lymphoid cells show 46,X,fra(X)(q27); proband is GM5131 Fibroblast
GM5855		R1640 15I	0148	24	F	WH	+-(O)	A	MOTHER	819	5	Two affected sons; one affected brother; mild speech difficulties; proband is GM5131 Fibroblast; 46,X,fra(X)(q27) in 6% of lymphoid cells
GM6912		R1640 15I	0061	17	M	BL	Y-		PROBAND	970	1	Mental retardation and hyperactivity; three affected male and one affected female cousins; positive for fra(X) in peripheral blood lymphocytes
GM6913		R1640 15I	0061	40	F	BL	+-(O)		MOTHER	970	2	One affected son; three affected nephews; one affected niece; sister of GM6863; negative for fra(X) in peripheral blood lymphocytes
GM6863		R1640 15I	0061	32	F	BL	+-(O)		MAT AUNT	970	3	Average IQ; one affected son; three affected nephews; one affected niece; negative for fra(X) in peripheral blood lymphocytes
GM6864		R1640 15I	0061	10	F	BL	+?		MAT COU	970	4	Normal IQ and development; one affected brother; four affected cousins; possible heterozygote; daughter of GM6863; negative for fra(X) in peripheral blood lymphocytes

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS											
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955											
GM6865		R1640 15I	0061	9	M	BL	++	MAT COU	970	5	Normal functioning and development; one affected brother; four affected cousins; son of GM6863; negative for fra(X) in peripheral blood lymphocytes
GM7063		R1640 15I	0061	16	F	BL	+-	AFF MAT COU	970	6	Slow learner; possible affected heterozygote 14/50 blood lymphocytes showed fra(X); four affected cousins; daughter of GM7064
GM7064		R1640 15I	0061	39	F	BL	+-(O)	MAT AUNT	970	7	Normal IQ; four sisters are carriers; non-expressing obligate carrier; one affected daughter; four affected nephews; negative for fra(X) in peripheral blood lymphocytes
GM7292		R1640 15I	0061	18	F	BL	+?	COU	970	8	Normal IQ; possible heterozygote; daughter of GM7064; 46,XX in peripheral blood lymphocytes
GM6897		R1640 15I	0061	25	M	WH	Y-	PROBAND	971	1	Mental retardation; typical facies; large ears; macro-orchidism; 46,fra(X)(q27),Y in peripheral blood lymphocytes
GM6894		R1640 15I	0061	50	F	WH	+-(O)	MOTHER	971	2	Clinically normal; one affected son and grandson; 46,XX in peripheral blood lymphocytes
GM6895		R1640 15I	0061	55	M	WH	++	FATHER	971	3	Clinically normal; one affected son and grandson; 46,XY in peripheral blood lymphocytes
GM6889		R1640 15I	0061	14	F	WH	+?	SIS	971	4	Clinically normal; possible heterozygote; affected brother and nephew; 46,XX in peripheral blood lymphocytes
GM6890		R1640 15I	0061	19	M	WH	Y?	BRO	971	5	Clinically normal; affected brother and nephew; 46,XY in peripheral blood lymphocytes
GM6891		R1640 15I	0061	29	M	WH	Y?	BRO	971	6	Clinically normal; affected brother and nephew; negative for fra(X) in peripheral blood lymphocytes
GM6893		R1640 15I	0061	12	F	WH	+?	SIS	971	7	Clinically normal; possible heterozygote; affected brother and nephew; 46,XX in peripheral blood lymphocytes

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMMB	REMARKS
X CHROMOSOME MARKERS												
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955												
GM6896		P1640 15I	0061	27	F	WH	+-(0)		SIS	971	8	Clinically normal; one affected son and brother; negative for fra(X) in peripheral blood lymphocytes
GM6892		R1640 15I	0061	84	M	WH	Y?		MAT GR FATH	971	9	Clinically normal; affected grandson and great-grandson; suspected non-expressing "carrier"; 46,XY in peripheral blood lymphocytes
GM6905		R1640 15I	0061	72	F	WH	+-(0)		MAT COU	971	10	Clinically normal; one affected son and great-grandson; negative for fra(X) in peripheral blood lymphocytes
GM6906		R1640 15I	0061	52	M	WH	Y?		MAT COU	971	11	Clinically normal; affected brother and grandson; son of GM6905; husband of GM6904; father of GM6903 & GM6907; suspected "carrier"; negative for fra(X) in peripheral blood lymphocytes
GM6904		R1640 15I	0061	52	F	WH	++		IN-LAW	971	12	Clinically normal; spouse of GM6906; mother of GM6903 & GM6907; one affected grandson; negative for fra(X) in peripheral blood lymphocytes
GM6903		R1640 15I	0061	30	F	WH	+-(0)		MAT COU	971	13	Clinically normal; affected son is GM6852; daughter of GM6904 & GM6906; negative for fra(X) in peripheral blood lymphocytes
GM6907		R1640 15I	0061	27	F	WH	+?		MAT COU	971	14	Clinically normal; affected nephew and uncle; possible heterozygote; sister of GM6903; daughter of GM6904 & GM6906; negative for fra(X) in peripheral blood lymphocytes
GM6852		R1640 15I	0061	5	M	WH	Y-		AFF MAT COU	971	15	Seizures; borderline IQ; large ears; positive family history for fra(X) and mental retardation; 46+fra(X)(q27)+Y in peripheral blood lymphocytes; son of GM6903
GM6911		R1640 15I	0061	27	F	WH	+?		MAT COU	971	16	Slow; probably clinically affected; daughter of GM6910; negative for fra(X) in peripheral blood lymphocytes
GM6910		R1640 15I	0061	55	F	WH	+?		MAT AUNT	971	17	Clinically normal; possible heterozygote; negative for fra(X) in peripheral blood lymphocytes

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
			X CHROMOSOME MARKERS								
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955											
GM7174		R1640 15I	0061	21	M	WH	Y?	BRO	971	18	46,XY in peripheral blood lymphocytes; could be a "non-expressing male" based upon family history; affected brother and nephew
GM7175		R1640 15I	0061	26	F	WH	+?	SIS	971	19	Clinically normal; 46,XX in peripheral blood lymphocytes; affected brother and nephew
GM7365		R1640 15I	0061	35	M	WH	Y-	AFF MAT COU	971	20	Mental retardation; macro-orchidism; staccato, unclear speech; positive for fra(X) in peripheral blood lymphocytes
GM7536		R1640 15I	0061	56	M	WH	Y?	MAT COU	971	22	Clinically normal; possible normal "carrier"; 46,XY in peripheral blood lymphocytes
GM7537		R1640 15I	0061	64	F	WH	+-(D)	MAT COU	971	23	Clinically normal; sister of GM7536; one affected son and two affected grandsons; positive for fra(X) in peripheral blood lymphocytes
GM7538		R1640 15I	0061	43	F	WH	+?	MAT COU	971	24	Clinically normal; possible heterozygote; one affected brother; daughter of GM7537; negative for fra(X) in peripheral blood lymphocytes
GM7539		R1640 15I	0061	66	M	WH	Y?	MAT COU	971	25	Clinically normal; possible normal "carrier"; brother of GM7536; 46,XY in peripheral blood lymphocytes
GM7540		R1640 15I	0061	55	F	WH	+?	MAT COU	971	26	Clinically normal; possible heterozygote; sister of GM7536; negative for fra(X) in peripheral blood lymphocytes
GM7542		R1640 15I	0061	56	M	WH	Y?	MAT COU	971	27	Clinically normal; twin of GM7536; possible normal "carrier"; 46,XY in peripheral blood lymphocytes
GM7541		R1640 15I	0061	37	F	WH	+?	MAT COU	971	28	Clinically normal; daughter of GM7540 who has a 50% risk of being a carrier; negative for fra(X) in peripheral blood lymphocytes
GM7543		R1640 15I	0061	24	F	WH	+?	MAT COU	971	29	Clinically normal; at least three sibs with mental retardation; mother is sister of GM7536; negative for fra(X) in peripheral blood lymphocytes

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
X CHROMOSOME MARKERS											
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955											
GM6968		R1640 15I	0061	32	F	WH	+-(O)	MOTHER	987	2	Obligate carrier; clinically normal; 1% of lymphocytes show fra(X) with FudR or MTX; affected uncle and nephew; father is a normal "carrier"; son is positive for fra(X); Proband is GM7071 Fibroblast
GM7131		R1640 15I	0061	63	M	WH	Y-	GR FATHER	987	3	Phenotypically normal "carrier"; 46,XY in 250 peripheral blood lymphocytes; father of GM6968 & GM7132
GM7132		R1640 15I	0061	35	F	WH	+-(O)	MAT AUNT	987	4	Clinically normal; one affected son and paternal uncle; sister of GM6968; 46,XX in peripheral blood lymphocytes
GM7294		R1640 15I	0061	23	M	BL	Y-	PROBAND	1011	2	Institutionalized for mental retardation; 46,fra(X)(q27),Y in 32% of leukocytes; G6PD type A+; a twin, not identical by HLA typing, is institutionalized but not known if mentally retarded
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890											
*GM0717A	30HR	R1640 15I	0066	11	M		--	PROBAND			Exhibits increased chromosomal radiation sensitivity; increased chromosome breakage induced by bleomycin
*GM0719	21HR	R1640 20I	0066	13	F	WH	--	PROBAND	177	1	46,XX; two sibs also affected; lymphoid culture shows hypersensitivity to X-irradiation
*GM0718	29HR	R1640 20I	0066	9	M	WH	--	AFF BRO	177	2	Two sibs also affected
*GM0736	44HR	R1640 20I	0066		M	WH	+-(O)	FATHER	177	3	46,XY; 3 affected children; clinically unaffected
*GM0781	67HR	R1640 20I	0066	4	F	WH	--	AFF SIS	177	4	Two sibs also affected
*GM1526A	35HR	R1640 15I	0054	28	F	WH	--	PROBAND	178	1	46,XX,t(X;14) clone in short term lymphocytes; HLA type: A9,A10,Bw17,B13(Bw40); AT8BI; exhibits increased chromosomal radiation sensitivity; similarly affected sib; increased chromosome breakage induced by bleomycin; 46,XX in lymp cult
*GM1525C		R1640 15I	0054	19	F	WH	--	AFF SIS	178	2	46,XX,t(14;14) clone in short term lymphocytes; 46,XX in lymphoid culture; HLA type A28,(Aw31,33),B12; AT2BI; exhibits increased chromosomal radiation sensitivity; increased chromosome breakage induced by bleomycin

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE											
Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890											
*GM2782	37HR	R1640 20I	0077	5	M	WH	--	PROBAND	228	1	Iranian; same patient as GM2532; similarly affected sib; see GM2531 Fibroblast; AT20IJE-F
*GM2783A	23HR	R1640 15I	0077	8	M	WH	--	AFF BRO	228	2	Iranian; AT21IJE-F; see GM2530 Fibroblast; hypersensitive to the lethal effects of gamma-irradiation
GM2781A		R1640 20I	0077	40	F	WH	+-(0)	MOTHER	228	3	Iranian; clinically normal; two affected cnildren
*GM3187	39HR	R1640 20I	0226	40	M	WH	+-(0)	FATHER	516	1	Clinically normal; ATH158e; two affected children; see GM3488 Fibroblast
*GM3188	16HR	R1640 20I	0226	36	F	WH	+-(0)	MOTHER	516	2	Clinically normal; ATH168e; two affected children; see GM3489 Fibroblast
*GM3189	19HR	R1640 20I	0226	7	M	WH	--	PROBAND	516	3	Neurological abnormalities; AT48e; similarly affected sib; see GM3487B Fibroblast; lymphoid culture shows hypersensitivity to X-irradiation
GM3324	23HR	R1640 20I	0226	15	F	WH	+?	SIS	516	4	Possible heterozygote; clinically normal; see GM3490 Fibroblast
GM3323	23HR	R1640 20I	0226	6	M	WH	+?	BRO	516	5	Possible heterozygote; clinically normal; see GM3492 Fibroblast
GM3325	23HR	R1640 20I	0226	14	F	WH	+?	SIS	516	6	Possible heterozygote; clinically normal; see GM3491 Fibroblast
GM3332	26HR	R1640 20I	0226	13	M	BL	--	PROBAND	605	1	See GM3395A Fibroblast; a deceased sister had A.T.; lymphoid culture shows hypersensitivity to X-irradiation
GM3382	18HR	R1640 20I	0226	43	M	BL	+-(0)	FATHER	605	2	See GM3397 Fibroblast; clinically normal; two affected children
GM3334	25HR	R1640 20I	0226	42	F	BL	+-(0)	MOTHER	605	3	See GM3396 Fibroblast; clinically normal; two affected children
GM3333	25HR	R1640 20I	0226	11	F	BL	+?	SIS	605	4	Normal; possible heterozygote; see GM3400 Fibroblast
GM3380	17HR	R1640 20I	0226	20	F	BL	+?	SIS	605	5	Normal; possible heterozygote; see GM3399 Fibroblast
GM3383	39HR	R1640 20I	0226	22	M	BL	+?	BRO	605	6	Normal; possible heterozygote; see GM3398 Fibroblast

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE												
Bloom Syndrome - 21090												
*GM3403C		R1640 15I	0226	33	M	WH	--	A	PROBAND	210	1	Ashkenazi; B.S. Registry #9(EmSh); telangiectasias & pigmentation; congenital dwarf; 46,XY; 40% of cells show random chromosome loss/gain; increased sister chromatid exchange; 8% of cells show breakage or fragments; see GM3402 Fibro.
*GM4408	20HR	R1640 15I	0224	4	M	WH	--		PROBAND	720	1	B.S. Registry #87(AlFr); 46,XY; excessive SCEs in leuko. & fibro.; norm. SCEs in lymphoid culture; 16% of cells show random chromo. loss; neg. for T cell 65,000 M.W. glycoprotein; pos. for sIg & Ia (B cell) antigen; see GM3498B Fibro.
Fanconi Anemia (Pancytopenia) - 22765, 22766												
*GM4510A		R1640 15I	0077	5	F	WH	--		PROBAND	734	1	Pancytopenia and digital anomalies; 45, XX,t(13;14)(13qter>cen>14qter); 24% of cells show 7p-; exhibits increased chromosome breakage induced by diepoxybutane, mitomycin-C, nitrogen mustard, and bleomycin
EXTENDED FAMILIES - LYMPHOBLAST CULTURES												
UTAH PEDIGREE K-1329 (See Appendix for Pedigree)												
GM6986		R1640 15U	0479	74	M	WH			I-1	981	1	Father of GM6997
GM7045		R1640 15U	0479	67	F	WH			I-2	981	2	Mother of GM6997
GM7051B		R1640 15U	0479	67	M	WH			I-3	981	3	Father of GM7014 & GM7013
GM7031		R1640 15U	0479	63	F	WH			I-4	981	4	Mother of GM7014 & GM7013
GM7435		R1640 15U	0479	77	M	WH			I-5	981	5	Father of GM6995, GM7042, GM7046, GM7434
GM7037		R1640 15U	0479	65	F	WH			I-6	981	6	Mother of GM6995, GM7042, GM7046, GM7434
GM6984A		R1640 15U	0479	63	M	WH			I-9	981	7	Father of GM7041
GM6989		R1640 15U	0479	62	F	WH			I-10	981	8	Mother of GM7041
GM6995		R1640 15U	0479	45	M	WH			II-1	981	9	Husband of GM6997
GM6997		R1640 15U	0479	40	F	WH			II-2	981	10	Wife of GM6995
GM7042		R1640 15U	0479	44	M	WH			II-3	981	11	Husband of GM7014

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
EXTENDED FAMILIES - LYMPHOBLAST CULTURES											
UTAH PEDIGREE K-1329 (See Appendix for Pedigree)											
GM7014		R1640 15U	0479	39	F	WH		II-4	981	12	Wife of GM7042
GM7046		R1640 15U	0479	39	M	WH		II-7	981	13	Husband of GM7041
GM7041		R1640 15U	0479	38	F	WH		II-8	981	14	Wife of GM7046
GM7434		R1640 15U	0479	38	F	WH		II-9	981	15	Wife of GM7013
GM7013		R1640 15U	0479	43	M	WH		II-10	981	16	Husband of GM7434
GM7018		R1640 15U	0479	18	M	WH		III-1	981	17	Son of GM6995
GM7036		R1640 15U	0479	17	M	WH		III-2	981	18	Son of GM6995
GM6981		R1640 15U	0479	15	F	WH		III-3	981	19	Daughter of GM6995
GM6980		R1640 15U	0479	13	F	WH		III-4	981	20	Daughter of GM6995
GM7047		R1640 15U	0479	11	M	WH		III-5	981	21	Son of GM6995
GM7433		R1640 15U	0479		M	WH		III-6	981	22	Son of GM6995
GM7058B		R1640 15U	0479	7	F	WH		III-7	981	23	Daughter of GM6995
GM7003		R1640 15U	0479	19	F	WH		III-9	981	24	Daughter of GM7042
GM7436		R1640 15U	0479	18	F	WH		III-10	981	25	Daughter of GM7042
GM7694		R1640 15U	0479	17	F	WH		III-11	981	26	Daughter of GM7042
GM6998		R1640 15U	0479	13	M	WH		III-12	981	27	Son of GM7042
GM7032		R1640 15U	0479	11	M	WH		III-13	981	28	Son of GM7042
GM7001A		R1640 15U	0479	10	M	WH		III-14	981	29	Son of GM7042
GM7431		R1640 15U	0479	9	M	WH		III-15	981	30	Son of GM7042
GM7028		R1640 15U	0479	16	M	WH		III-21	981	31	Son of GM7046
GM7039		R1640 15U	0479	15	M	WH		III-22	981	32	Son of GM7046
GM7025		R1640 15U	0479	13	F	WH		III-23	981	33	Daughter of GM7046
GM7437		R1640 15U	0479	12	F	WH		III-24	981	34	Daughter of GM7046
GM7060		R1640 15U	0479	11	M	WH		III-25	981	35	Son of GM7046

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
EXTENDED FAMILIES - LYMPHOBLAST CULTURES											
UTAH PEDIGREE K-1329 (See Appendix for Pedigree)											
GM7061A		R1640 15U	0479	8	M	WH		III-26	981	36	Son of GM7046
GM7432		R1640 15U	0479	6	M	WH		III-27	981	37	Son of GM7046
GM7035		R1640 15U	0479	18	M	WH		III-31	981	38	Son of GM7013
GM7054		R1640 15U	0479	17	M	WH		III-32	981	39	Son of GM7013
GM6996		R1640 15U	0479	15	F	WH		III-33	981	40	Daughter of GM7013
GM7015		R1640 15U	0479	13	M	WH		III-34	981	41	Son of GM7013
GM7024		R1640 15U	0479	11	M	WH		III-35	981	42	Son of GM7013
GM7043A		R1640 15U	0479	9	M	WH		III-37	981	43	Son of GM7013
UTAH PEDIGREE K-1331 (See Appendix for Pedigree)											
GM7050A		R1640 15U	0479	62	F	WH		I-1	982	1	Mother of GM6990
GM7016		R1640 15U	0479	71	M	WH		I-2	982	2	Father of GM6990
GM7340A		R1640 15U	0479	83	F	WH		I-3	982	3	Mother of GM7057
GM7007		R1640 15U	0479	95	M	WH		I-4	982	4	Father of GM7057
GM6990		R1640 15U	0479	41	F	WH		II-1	982	5	Wife of GM7057
GM7057		R1640 15U	0479	48	M	WH		II-2	982	6	Husband of GM6990
GM7023		R1640 15U	0479	21	F	WH		III-1	982	7	Daughter of GM6990
GM7033		R1640 15U	0479	20	M	WH		III-2	982	8	Son of GM6990
GM7059		R1640 15U	0479	18	F	WH		III-3	982	9	Daughter of GM6990
GM7005		R1640 15U	0479	16	F	WH		III-4	982	10	Daughter of GM6990
GM6999		R1640 15U	0479	15	F	WH		III-5	982	11	Daughter of GM6990
GM6988		R1640 15U	0479	13	F	WH		III-6	982	12	Daughter of GM6990
GM6983		R1640 15U	0479	11	M	WH		III-7	982	13	Son of GM6990
GM7030		R1640 15U	0479	9	M	WH		III-8	982	14	Son of GM6990
GM6992		R1640 15U	0479	8	M	WH		III-9	982	15	Son of GM6990
UTAH PEDIGREE K-1333 (See Appendix for Pedigree)											
*GM7341		R1640 15U	0479	61	F	WH		I-1	983	1	Mother of GM6987A

BIOCHEMICALLY MUTANT HUMAN LYMPHOBLASTS

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
EXTENDED FAMILIES - LYMPHOBLAST CULTURES											
UTAH PEDIGREE K-1333 (See Appendix for Pedigree)											
*GM7017		R1640 15U	0479	61	M	WH		I-2	983	2	Father of GM6987A
*GM7002		R1640 15U	0479	63	F	WH		I-3	983	3	Mother of GM7038A
*GM7049		R1640 15U	0479	68	M	WH		I-4	983	4	Father of GM7038A
*GM6987A		R1640 15U	0479	39	F	WH		II-1	983	5	Wife of GM7038A
*GM7038A		R1640 15U	0479	41	M	WH		II-2	983	6	Husband of GM6987A
*GM7004		R1640 15U	0479	19	M	WH		III-1	983	7	Son of GM6987A
*GM7052		R1640 15U	0479	17	M	WH		III-2	983	8	Son of GM6987A
*GM6982		R1640 15U	0479	16	M	WH		III-3	983	9	Son of GM6987A
*GM7011		R1640 15U	0479	14	F	WH		III-4	983	10	Daughter of GM6987A
*GM7009		R1640 15U	0479	12	M	WH		III-5	983	11	Son of GM6987A
*GM7678		R1640 15U	0479	12	M	WH		III-6	983	12	Son of GM6987A
*GM7026		R1640 15U	0479	6	M	WH		III-7	983	13	Son of GM6987A
*GM7679		R1640 15U	0479	6	M	WH		III-8	983	14	Son of GM6987A
UTAH PEDIGREE K-1340 (See Appendix for Pedigree)											
GM7056		R1640 15U	0479	65	F	WH		I-1	984	1	Mother of GM7019
GM7022		R1640 15U	0479	63	M	WH		I-2	984	2	Father of GM7019
GM7000		R1640 15U	0479	66	F	WH		I-3	984	3	Mother of GM7029
GM6994		R1640 15U	0479	68	M	WH		I-4	984	4	Father of GM7029
GM7019		R1640 15U	0479	45	F	WH		II-1	984	5	Wife of GM7029
GM7029		R1640 15U	0479	47	M	WH		II-2	984	6	Husband of GM7019
GM7062		R1640 15U	0479	27	F	WH		III-1	984	7	Daughter of GM7019
GM7053A		R1640 15U	0479	24	F	WH		III-2	984	8	Daughter of GM7019
GM7008		R1640 15U	0479	23	M	WH		III-3	984	9	Son of GM7019
GM7040		R1640 15U	0479	19	M	WH		III-4	984	10	Son of GM7019
GM7342		R1640 15U	0479	16	M	WH		III-5	984	11	Son of GM7019

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
EXTENDED FAMILIES - LYMPHOBLAST CULTURES											
UTAH PEDIGREE K-1340 (See Appendix for Pedigree)											
GM7027		R1640 15U	0479	11	M	WH		III-6	984	12	Son of GM7019
UTAH PEDIGREE K-1341 (See Appendix for Pedigree)											
GM6985A		R1640 15U	0479	69	F	WH		I-1	985	1	Mother of GM6991
GM6993		R1640 15U	0479	74	M	WH		I-2	985	2	Father of GM6991
GM7055A		R1640 15U	0479	70	F	WH		I-3	985	3	Mother of GM7048
GM7034A		R1640 15U	0479	71	M	WH		I-4	985	4	Father of GM7048
GM6991		R1640 15U	0479	42	F	WH		II-1	985	5	Wife of GM7048
GM7048		R1640 15U	0479	43	M	WH		II-2	985	6	Husband of GM6991
GM7343		R1640 15U	0479	22	F	WH		III-1	985	7	Daughter of GM6991
GM7044		R1640 15U	0479	20	F	WH		III-2	985	8	Daughter of GM6991
GM7012		R1640 15U	0479	18	F	WH		III-3	985	9	Daughter of GM6991
GM7344		R1640 15U	0479	17	F	WH		III-4	985	10	Daughter of GM6991
GM7021		R1640 15U	0479	14	M	WH		III-5	985	11	Son of GM6991
GM7006		R1640 15U	0479	10	F	WH		III-6	985	12	Daughter of GM6991
GM7010A		R1640 15U	0479	8	F	WH		III-7	985	13	Daughter of GM6991
GM7020		R1640 15U	0479	7	M	WH		III-8	985	14	Son of GM6991
UTAH PEDIGREE K-1345 (See Appendix for Pedigree)											
GM7345		R1640 15U	0479	69	F	WH		I-1	1029	1	Mother of GM7348A
GM7357		R1640 15U	0479	69	M	WH		I-2	1029	2	Father of GM7348A
GM7346		R1640 15U	0479	85	F	WH		I-3	1029	3	Mother of GM7349
GM7347		R1640 15U	0479	86	M	WH		I-4	1029	4	Father of GM7349
GM7348A		R1640 15U	0479	45	F	WH		II-1	1029	5	Wife of GM7349
GM7349		R1640 15U	0479	49	M	WH		II-2	1029	6	Husband of GM7348A
GM7350		R1640 15U	0479	25	F	WH		III-1	1029	7	Daughter of GM7348A
GM7351		R1640 15U	0479	23	M	WH		III-2	1029	8	Son of GM7348A

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REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
EXTENDED FAMILIES - LYMPHOBLAST CULTURES												
UTAH PEDIGREE K-1345 (See Appendix for Pedigree)												
GM7352		R1640 15U	0479	22	M	WH			III-3	1029	9	Son of GM7348A
GM7353A		R1640 15U	0479	20	M	WH			III-4	1029	10	Son of GM7348A
GM7354		R1640 15U	0479	19	F	WH			III-5	1029	11	Daughter of GM7348A
GM7355A		R1640 15U	0479	16	M	WH			III-6	1029	12	Son of GM7348A
GM7356		R1640 15U	0479	13	M	WH			III-7	1029	13	Son of GM7348A
SPECIALLY CHARACTERIZED HUMAN LYMPHOBLAST CULTURES												
HLA Histocompatability Type - 14280 - 14286												
*GM3098A	23HR	R1640 20I	0300		F		A		PROBAND			HLA TYPE: A:2,2;B:17,17;DR:3,3 Bodmer I.D.: WT49; 46,XX; 10% of cells show random chromosome loss/gain; consanguineous parents
*GM3099	43HR	R1640 20I	0300		F		A		PROBAND			HLA type; A:2,-,B:35,-,C:4,-,D:1,1, DR:1,1; Bodmer I.D.: MAJA; 46,XX
*GM3103A		R1640 15I	0300		M		A		PROBAND			HLA type; A:9,9,B:14,14,DR:4,4 Bodmer I.D.: WT51; 46,XY; consanguineous parents
*GM3104A	22HR	R1640 20I	0300				A		PROBAND			HLA type; A:3,3,B:35,35,C:4,4,D:1,1, DR:1,1; Bodmer I.D.: IBW4; consanguineous parents
GM3105A	29HR	R1640 20I	0300				A		PROBAND			HLA type; A:26,28,B:38,18,D:5,DR:5 Bodmer I.D.: IDF
*GM3106	31HR	R1640 20I	0300				A		PROBAND			HLA type; A:1,B:35,C:4,DR:5,DC:1 Bodmer I.D.: FPF; consanguineous parents
*GM3107	31HR	R1640 20I	0300				A		PROBAND			HLA type; A:3,B:7,D:2,DR:2 Bodmer I.D.: PGF; consanguineous parents
GM3153A		R1640 15I	0300		M		A		PROBAND			HLA type; A:2,2,B:38,39,DR:6 Bodmer I.D.: ARENT; 46,XY
*GM3160	20HR	R1640 20I	0300				A		PROBAND			HLA type; A:2,2,B:35,12,DR:4x7 Bodmer I.D.: HERLUFF
*GM3161	17HR	R1640 20I	0300				A		PROBAND			HLA type; A:3,3,B:7,7,D:2,2,DR:2,2 Bodmer I.D.: MST1
*GM3162	18HR	R1640 20I	0300				A		PROBAND			HLA type; A:2,B:8,35,D:3,DR:3 Bodmer I.D.: MWF

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SPECIALLY CHARACTERIZED HUMAN LYMPHOBLAST CULTURES											
HLA Histocompatability Type - 14280 - 14286											
*GM3163	30HR	R1640 20I	0300				A	PROBAND			HLA type: A:30,B:13,C:w6,DR:7 Bodmer I.D.: LBF; consanguineous parents
*GM3164	24HR	R1640 20I	0300				A	PROBAND			HLA type: A:2,24,B:27,C:2,D:4,DR:4,4 Bodmer I.D.: AS
*GM3190	23HR	R1640 20I	0300	17	M	BL	A	PROBAND			B cell line, no HLA: A, B, or C; no B2 Microglobulin; HLA: D:6; Bodmer I.D.: DAUDI; culture is negative for T cell 65,000 M.W. glycoprotein; positive for sIg and Ia (B cell) antigen; weak response to anti-HLA antibody
*GM3357A	23HR	R1640 15I	0300				A	PROBAND			HLA type: A:1,2,B:8,13,C:w6,w7,DR:2 Bodmer I.D.: BRISTOL-8
*GM6815		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: Aw24,Bw60,Cw3,Dw9, DRw9,& SB4; product of consanguineous (first cousin) marriage; line DKB
GM6816		R1640 15I	0463		F	WH	B	PROBAND			Homozygous HLA type: A2,Bw38,Dw6,DRw6,& SB2,4; product of consanguineous (first cousin) marriage; line WVD
*GM6817		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A1,Bw60,Dw6,DRw6,& SB"4"; product of consanguineous (first cousin) marriage; line APD
*GM6818		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A3,B7,Cw7,Dw6,DRw6, & SB4; product of consanguineous (first cousin) marriage; line HHK
*GM6819		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A2,B18,Cw5,Dw7,DR5, & SB2; product of consanguineous (first cousin) marriage; line JVM
*GM6820A		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A25,B18,Dw5,DR5,& SB2; product of consanguineous (first cousin) marriage; line THR
GM6821		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A2,Bw62,Cw3,Dw4, DR4,& SB2; product of consanguineous (first cousin) marriage; line BSM
*GM6822		R1640 15I	0463			WH	B	PROBAND			Homozygous HLA type: A1,B8,Cw7,Dw3,DR3,& SB4; product of consanguineous (first cousin) marriage; line HAR

REP #	GEN TIME	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
SPECIALLY CHARACTERIZED HUMAN LYMPHOBLAST CULTURES												
HLA Histocompatability Type - 14280 - 14286												
*GM6824		R1640 15I	0463			WH		B	PROBAND			Homozygous HLA type: A26,B18,Cw5,Dw3, DR3,6 SB2; product of consanguineous (first cousin) marriage; line QBL
GM6827		R1640 15I	0463			WH		B	PROBAND			Homozygous HLA type: A26,Bw56,Cw1,Dw1, DR1,6 SB2; product of consanguineous (first cousin) marriage; line QOS
*GM7254		R1640 15U	0493	4	M	WH			PROBAND			Consanguineous parents; cleft palate and renal agenesis; homozygous HLA type: A1, B39,Cw7,DRw6.1; BF type:1-1; GLO1 type: 2-2; GC type:2-1; ACP1 type:BB; ADA type:1-1; ADK type:1-1; ESD type:1-1; PGM1 type:1-1; HP type:1-1
*GM6823		R1640 15I	0463		F	WH		B	PROBAND	1052	1	Homozygous HLA type: A1,B8,Cw7,Dw3,DR3,6 SB2; product of consanguineous (first cousin) marriage; line AVL
GM6826		R1640 15I	0463		F	WH		B	SIS	1052	2	Homozygous HLA type: Aw32,B27,Cw2,Dw1, DR1,6 SB2; product of consanguineous (first cousin) marriage; line MVL

Chromosomally Aberrant Human Lymphoblasts

Chromosomally Aberrant Human Lymphoblasts

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							TRANSLOCATION						
CHROMOSOME X													
*GM6007		R1640	15I	0148	10	F	46,X,t(X;9)(Xqter>Xp21.2::9p22>9pter; 9qter>9p22::Xp21.2>Xpter)	B	A	PROBAND			Duchenne Muscular dystrophy; elevated CPK; EMG and muscle biopsy consistent with DMD; normal X is late replicating; parents both have normal karyotypes
*GM3316		R1640	20I	0311	3	F	WH 46,X,del(11)(p11.2p1300),t(X;11)(Xpter> Xq11.1::11p1300>11pter;11qter>11p11.2:: Xq11.1>Xqter)	U	A	PROBAND	617	1	South American; normal X is late replicating; mental retardation with bizarre dysmorphology syndrome; see GM2859 Fibroblast
*GM5011		R1640	15I	0398	19	F	WH 46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2> 2qter;2pter>2q14.2::8p11.1>8pter;8qter> 8p11.1::Xq21.2>Xqter)	B	A	PROPAND	808	1	Primary amenorrhea; lack of secondary sexual development; normal IQ; no dysmorphic features; see GM5010 Fibroblast; possible del(X)(q21.2q22.3); normal X is late replicating
CHROMOSOME Y													
*GM6326		R1640	15I	0455	34	M	WH 46,X,t(Y;17)(Ypter>Yq11.21::17q12> 17qter;17pter>17q12::Yq11.21>Yqter)	B	A	PROBAND			Infertility; azoospermia
CHROMOSOME 01													
GM3423		R1640	20I	0058	32	F	WH 46,XX,t(1;6)(1qter>1p36::6p23>6pter; 6qter>6p23::1p36>1pter)	B	A	PROBAND			Puerto Rican
GM6924		R1640	15I	0454	31	M	WH 46,XY,t(1;3)(1pter>1q32.1::3p25.1>3pter; 3qter>3p25.1::1q32.1>1qter)	B	A	PROBAND			Clinically normal
GM5348		R1640	15I	0074	29	F	WH 46,XX,t(1;9)(1pter>1q32::9p24>9pter; 9qter>9p24::1q32>1qter)	B	A	MOTHER	849	2	Son is GM5347 Fibroblast
GM7158		R1640	15I	0491	2	M	OT 46,XY,-2,+der(2),t(1;2)(2pter>2q37:: 1q32.1>1qter)mat	U	A	PROBAND	876	1	Severely retarded and dysmorphic; Fumarate hydratase activity consistent with trisomy for 1q
GM6226		R1640	15I	0148	6	M	WH 46,XY,-1,+der(1),t(1;16)(1pter>1q44:: 16p13.11>16pter)mat	U	A	PROBAND	932	1	Hb alpha/Hb beta ratio equals 3:2 in RBC
GM6227		R1640	15I	0148	33	F	WH 46,XX,t(1;16)(1pter>1q44::16p13.11> 16pter;16qter>16p13.11::1q44>1qter)	B	A	MOTHER	932	2	Clinically normal; Hb alpha/Hb beta ratio equals 1:1 in RBC
CHROMOSOME 02													
GM3559		R1640	20I	0328	30	M	WH 46,XY,t(2;15)(2pter>2q37::15q22>15qter; 15pter>15q22::2q37>2qter)	B	A	PROBAND			

CHROMOSOMALLY ABERRANT HUMAN LYMPHOBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 02													
*GM5011		R1640	15I	0398	19	F	WH 46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2> 2qter;2pter>2q14.2::8p11.1>8pter;8qter> 8p11.1::Xq21.2>Xqter)	B	A	PROBAND	808	1	Primary amenorrhea; lack of secondary sexual development; normal IQ; no dysmorphic features; see GM5010 Fibroblast; possible del(X)(q21.2q22.3); normal X is late replicating
GM7158		R1640	15I	0491	2	M	OT 46,XY,-2,+der(2),t(1;2)(2pter>2q37:: 1q32.1>1qter)mat	U	A	PROBAND	876	1	Severely retarded and dysmorphic; Fumarate hydratase activity consistent with trisomy for 1q
CHROMOSOME 03													
GM6924		R1640	15I	0454	31	M	WH 46,XY,t(1;3)(1pter>1q32.1::3p25.1>3pter; 3qter>3p25.1::1q32.1>1qter)	B	A	PROBAND			Clinically normal
CHROMOSOME 04													
GM1203A		R1640	15I	0066	30	F	WH 46,XX,t(4;12)(4pter>4q25::12p13>12pter; 12qter>12p13::4q25>4qter)	B	A	PROBAND			
GM3799		R1640	20I	0067	27	F	WH 46,XX,t(4;11)(4pter>4q34::11p11>11pter; 11qter>11p11::4q34>4qter)	B	A	PROBAND			Clinically normal
GM1561		R1640	20I	0008	36	F	WH 46,XX,t(4;11)(4pter>4q23::11q11>11qter; 11pter>11q11::4q23>4qter)	B	A	PARENT	397	1	27% of cells have 19p+; HLA type A10, Aw24,Bw35,(B12); see GM0380 Fibroblast
*GM1063A		R1640	20I	0067	12	F	BL 46,XX,t(4;13)(4pter>4q31::13q14>13qter; 13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Karyotype unstable; HLA type A1,B12, Bw17; see GM0972 Fibroblast
GM5654		R1640	15I	0354	40	F	WH 46,XX,t(4;11)(4pter>4q25::11p13>11pter; 11qter>11p13::4q25>4qter)	B	A	MOTHER	857	2	Clinically normal; son is GM5519 Fibroblast
CHROMOSOME 05													
*GM7173		R1640	15I	0107	22	M	WH 46,XY,t(5;7)(5pter>5q35::7q22>7qter; 7pter>7q22::5q35>5qter)mat	B	A	PROBAND			Balanced carrier
CHROMOSOME 06													
*GM2669		R1640	20I	0039	27	M	WH 46,XY,t(6;8)(6pter>6q27::8q21>8qter; 8pter>8q21::6q27>6qter)	B	A	PROBAND			Clinically normal
GM3423		R1640	20I	0058	32	F	WH 46,XX,t(1;6)(1qter>1p36::6p23>6pter; 6qter>6p23::1p36>1pter)	B	A	PROBAND			Puerto Rican
GM5184		R1640	15I	0039	49	M	WH 46,XY,t(6;7)(6pter>6q21::7q21.2>7qter; 7pter>7q21.2::6q21>6qter)	B	A	PROBAND	827	1	Wife has had three first trimester spontaneous abortions; see GM5183A Fibroblast

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 07													
*GM0633		R1640	20I	0089	8	M	WH 46,XY,t(7;10)(7qter>7p21::10q11>10qter; 10pter>10q11::7p21>7pter)	B	A	PROBAND			
*GM7173		R1640	15I	0107	22	M	WH 46,XY,t(5;7)(5pter>5q35::7q22>7qter; 7pter>7q22::5q35>5qter)mat	B	A	PROBAND			Balanced carrier
GM5184		R1640	15I	0039	49	M	WH 46,XY,t(6;7)(6pter>6q21::7q21.2>7qter; 7pter>7q21.2::6q21>6qter)	B	A	PROBAND	827	1	Wife has had three first trimester spontaneous abortions; see GM5183A Fibroblast
CHROMOSOME 08													
*GM2669		R1640	20I	0039	27	M	WH 46,XY,t(6;8)(6pter>6q27::8q21>8qter; 8pter>8q21::6q27>6qter)	B	A	PROBAND			Clinically normal
GM6832		R1640	15I	0148	29	F	WH 46,XX,t(8;15)(8qter>8p23::15q25>15qter; 15pter>15q25::8p23>8pter)	B	A	PROBAND			
*GM1921A		R1640	15I	0061	23	M	WH 47,XY,+21,inv(9)(pter>p11::q13>p11::q13> qter)mat,t(8;14)(8pter>8q11::14q11> 14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	See GM1920 Fibroblast; Down's Syndrome phenotype
*GM5011		R1640	15I	0398	19	F	WH 46,X,t(X;2;8)(Xpter>Xq21.2::2q14.2> 2qter;2pter>2q14.2::8p11.1>8pter;8qter> 8p11.1::Xq21.2>Xqter)	B	A	PROBAND	808	1	Primary amenorrhea; lack of secondary sexual development; normal IQ; no dysmorphic features; see GM5010 Fibroblast; possible del(X)(q21.2q22.3); normal X is late replicating
CHROMOSOME 09													
*GM6007		R1640	15I	0148	10	F	46,X,t(X;9)(Xqter>Xp21.2::9p22>9pter; 9qter>9p22::Xp21.2>Xpter)	B	A	PROBAND			Duchenne Muscular dystrophy; elevated CPK; EMG and muscle biopsy consistent with DMD; normal X is late replicating; parents both have normal karyotypes
GM2820		R1640	20I	0107	7MO	F	BL 46,XX,-12,+der(12),t(9;12)(12pter> 12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2819 Fibroblast
GM2822		R1640	20I	0107		F	BL 46,XX,t(9;12)(9qter>9p13::12q24>12qter; 12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2821 Fibroblast; balanced carrier
*GM1388		R1640	20I	0061	14	F	WH 46,XX,-13,+der(9),t(9;13)(9pter>9q22:: 13q12>13qter)mat or 46,XX,-13,+der(9), t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A	PROBAND	369	1	Profoundly retarded; clinical features of trisomy 9p syndrome; see GM1387 Fibroblast
GM5348		R1640	15I	0074	29	F	WH 46,XX,t(1;9)(1pter>1q32::9p24>9pter; 9qter>9p24::1q32>1qter)	B	A	MOTHER	849	2	Son is GM5347 Fibroblast

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 09													
GM7456		R1640	15I	0107	39	F	WH 46,XX,t(9;13)(9pter>9q31.2::13q32.1>13qter;13pter>13q32.1::9q31.2>9qter)	B	A	MOTHER	1042	2	Clinically normal; daughter is GM5319 Fibroblast
CHROMOSOME 10													
*GM0633		R1640	20I	0089	8	M	WH 46,XY,t(7;10)(7qter>7p21::10q11>10qter;10pter>10q11::7p21>7pter)	B	A	PROBAND			
GM5878		R1640	15I	0148	28	M	WH 46,XY,t(10;22)(10pter>10q26.3::22q11.2>22qter;22pter>22q11.2::10q26.3>10qter)	B	A	PARENT			Clinically normal; father of a child with DiGeorge Syndrome; proband not in Repository
GM6135		R1640	15I	0148	28	F	WH 46,XX,t(10;21)(10qter>10p11.2::21q22.3>21qter;21pter>21q22.3::10p11.2>10pter)	B	A	MOTHER	908	1	Clinically normal; balanced carrier
GM6136		R1640	15I	0148	2	M	WH 46,XY,-21,+der(21),t(10;21)(21pter>21q22.3::10p11.2>10pter)mat	U	A	PROBAND	908	2	Multiple congenital anomalies; bilateral cleft lip and palate; absent patellae; severe colconeo valgus and metatarsus abductus; mother is GM6135 Lymphoid
CHROMOSOME 11													
GM3799		R1640	20I	0067	27	F	WH 46,XX,t(4;11)(4pter>4q34::11p11>11pter;11qter>11p11::4q34>4qter)	B	A	PROBAND			Clinically normal
GM6275		R1640	15I	0148	29	F	WH 46,XX,t(11;22)(11pter>11q23::22q11>22qter;22pter>22q11::11q23>11qter)	B	A	PROBAND			One child has der(22) syndrome
GM1561		R1640	20I	0008	36	F	WH 46,XX,t(4;11)(4pter>4q23::11q11>11qter;11pter>11q11::4q23>4qter)	B	A	PARENT	397	1	27% of cells have 19p+; HLA type A10, Aw24,Bw35,(B12); see GM0380 Fibroblast
*GM3316		R1640	20I	0311	3	F	WH 46,X,del(11)(p11.2p1300),t(X;11)(Xpter>Xq11.1::11p1300>11pter;11qter>11p11.2::Xq11.1>Xqter)	U	A	PROBAND	617	1	South American; normal X is late replicating; mental retardation with bizarre dysmorphology syndrome; see GM2859 Fibroblast
GM5654		R1640	15I	0354	40	F	WH 46,XX,t(4;11)(4pter>4q25::11p13>11pter;11qter>11p13::4q25>4qter)	B	A	MOTHER	857	2	Clinically normal; son is GM5519 Fibroblast
GM6228		R1640	15I	0148	9MO	F	47,XX,+der(22),t(11;22)(22pter>22q11::11q23.3>11qter)mat	U	A	PROBAND	933	1	Hypotonia, developmental delay, ear pits, long philtrum, microretrognathia, high palate, long tapered fingers, slight frontal bossing, and small upslanted palpebral fissures; mother is GM6229 Lymphoid

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAM##	REMARKS
							TRANSLOCATION						
CHROMOSOME 11													
GM6229		R1640	15I	0148	23	F	WH 46,XX,t(11;22)(11pter>11q23.3::22q11>22qter;22pter>22q11::11q23.3>11qter)	B	A	MOTHER	933	2	Clinically normal; balanced carrier
GM6316A		R1640	15I	0148	35	F	WH 46,XX,t(11;16)(11qter>11p11.2::16p11.1>16pter;16qter>16p11.1::11p11.2>11pter)	B	A	PARENT	951	2	Mother of a child with an unbalanced translocation; husband is GM6315 Lymphoid
CHROMOSOME 12													
*GM0892A		R1640	20I	0024	12	F	WH 45,XX,-18,der(12),t(12;18)(12qter>12p11::18q11>18qter)/47,XX,+12	U	A	PROBAND			HLA type A2(A9),B12,B18; clinically normal; 22% of cells are 47,XX,+12
GM1203A		R1640	15I	0066	30	F	WH 46,XX,t(4;12)(4pter>4q25::12p13>12pter;12qter>12p13::4q25>4qter)	B	A	PROBAND			
GM2820		R1640	20I	0107	7MO	F	BL 46,XX,-12,+der(12),t(9;12)(12pter>12q24::9p13>9pter)mat	U	A	PROBAND	324	1	See GM2819 Fibroblast
GM2822		R1640	20I	0107		F	BL 46,XX,t(9;12)(9qter>9p13::12q24>12qter;12pter>12q24::9p13>9pter)	B	A	MOTHER	324	2	See GM2821 Fibroblast; balanced carrier
CHROMOSOME 13													
*GM1261		R1640	20I	0107	13	F	WH 45,XX,-13,-18,+t(13;18)(13qter>cen>18qter)	U	A	PROBAND			HLA type A1,A2,Bw15(B18); 18p- syndrome
*GM1388		R1640	20I	0061	14	F	WH 46,XX,-13,+der(9),t(9;13)(9pter>9q22::13q12>13qter)mat or 46,XX,-13,+der(9),t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A	PROBAND	369	1	Profoundly retarded; clinical features of trisomy 9p syndrome; see GM1387 Fibroblast
*GM2927		R1640	20I	0107	34	F	WH 46,XX,t(13;18)(13qter>13p13::18q122>18qter:18pter>18q122::13p13>13pter)	B	A	PROBAND	459	3	Clinically normal
*GM1063A		R1640	20I	0067	12	F	BL 46,XX,t(4;13)(4pter>4q31::13q14>13qter;13pter>13q14::4q31>4qter)	B	A	PROBAND	638	1	Karyotype unstable; HLA type A1,B12,Bw17; see GM0972 Fibroblast
*GM4510A		R1640	15I	0077	5	F	WH 45,XX,t(13;14)(13qter>cen>14qter)	U		PROBAND	734	1	Fanconi anemia; pancytopenia and digital anomalies; 24% of cells show 7p-; exhibits increased chromosome breakage induced by diepoxybutane, mitomycin-C, nitrogen mustard, and bleomycin
GM7456		R1640	15I	0107	39	F	WH 46,XX,t(9;13)(9pter>9q31.2::13q32.1>13qter;13pter>13q32.1::9q31.2>9qter)	B	A	MOTHER	1042	2	Clinically normal; daughter is GM5319 Fibroblast

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 14													
GM7912		R1640	15I	0016	38	M	WH 46,XY,t(14;17)(14pter>14q24.3::17p13.1>17pter;17qter>17p13.1::14q24.3>14qter)	B	A	PROBAND			Clinically normal
*GM1921A		R1640	15I	0061	23	M	WH 47,XY,+21,inv(9)(pter>p11::q13>p11::q13>qter)mat,t(8;14)(8pter>8q11::14q11>14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	See GM1920 Fibroblast; Down's Syndrome phenotype
*GM4510A		R1640	15I	0077	5	F	WH 45,XX,t(13;14)(13qter>cen>14qter)	U		PROBAND	734	1	Fanconi anemia: pancytopenia and digital anomalies; 24% of cells show 7p-; exhibits increased chromosome breakage induced by diepoxybutane, mitomycin-C, nitrogen mustard, and bleomycin
CHROMOSOME 15													
GM3559		R1640	20I	0328	30	M	WH 46,XY,t(2;15)(2pter>2q37::15q22>15qter;15pter>15q22::2q37>2qter)	B	A	PROBAND			
GM6832		R1640	15I	0148	29	F	WH 46,XX,t(8;15)(8qter>8p23::15q25>15qter;15pter>15q25::8p23>8pter)	B	A	PROBAND			
CHROMOSOME 16													
GM2324		R1640	20I	0182	27	F	WH 46,XX,t(16;22)(16qter>16p13::22q12>22qter;22pter>22q21::16p13>16pter),9qh+	B	A	MOTHER	179	2	Mother of GM2325 Fibroblast
GM6226		R1640	15I	0148	6	M	WH 46,XY,-1,+der(1),t(1;16)(1pter>1q44::16p13.11>16pter)mat	U	A	PROBAND	932	1	Hb alpha/Hb beta ratio equals 3:2 in RBC
GM6227		R1640	15I	0148	33	F	WH 46,XX,t(1;16)(1pter>1q44::16p13.11>16pter;16qter>16p13.11::1q44>1qter)	B	A	MOTHER	932	2	Clinically normal; Hb alpha/Hb beta ratio equals 1:1 in RBC
GM6316A		R1640	15I	0148	35	F	WH 46,XX,t(11;16)(11qter>11p11.2::16p11.1>16pter;16qter>16p11.1::11p11.2>11pter)	B	A	PARENT	951	2	Mother of a child with an unbalanced translocation; husband is GM6315 Lymphoid
CHROMOSOME 17													
*GM6326		R1640	15I	0455	34	M	WH 46,X,t(Y;17)(Ypter>Yq11.21::17q12>17qter;17pter>17q12::Yq11.21>Yqter)	B	A	PROBAND			Infertility; azoospermia
GM7912		R1640	15I	0016	38	M	WH 46,XY,t(14;17)(14pter>14q24.3::17p13.1>17pter;17qter>17p13.1::14q24.3>14qter)	B	A	PROBAND			Clinically normal
*GM3197		R1640	20I	0318	31	F	WH 46,XX,inv(5)(pter>p13::q13>p13::q13>qter),t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND	518	1	No hematological disorder; habitual aborter; normal phenotype; see GM3196 Fibroblast

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 18													
*GM0892A		R1640	20I	0024	12	F	WH 45,XX,-18,der(12),t(12;18)(12qter>12p11::18q11>18qter)/47,XX,+12	U	A	PROBAND			HLA type A2(A9),B12,B18; clinically normal; 22% of cells are 47,XX,+12
*GM1261		R1640	20I	0107	13	F	WH 45,XX,-13,-18,+t(13;18)(13qter>cen>18qter)	U	A	PROBAND			HLA type A1,A2,Bw15(B18); 18p- syndrome
*GM2927		R1640	20I	0107	34	F	WH 46,XX,t(13;18)(13qter>13p13::18q122>18qter;18pter>18q122::13p13>13pter)	B	A	PROBAND	459	3	Clinically normal
CHROMOSOME 21													
GM6135		R1640	15I	0148	28	F	WH 46,XX,t(10;21)(10qter>10p11.2::21q22.3>21qter;21pter>21q22.3::10p11.2>10pter)	B	A	MOTHER	908	1	Clinically normal; balanced carrier
GM6136		R1640	15I	0148	2	M	WH 46,XY,-21,+der(21),t(10;21)(21pter>21q22.3::10p11.2>10pter)mat	U	A	PROBAND	908	2	Multiple congenital anomalies; bilateral cleft lip and palate; absent patellae; severe colconeo valgus and metatarsus abductus; mother is GM6135 Lymphoid
GM6943		R1640	15I	0074	24	F	WH 46,XX,t(21;22)(21qter>21p12::22q11.1>22qter;22pter>22q11.1::21p12>21pter)	B	A	PARENT	977	1	Son has unbalanced translocation and Down's Syndrome; see GM6944 Fibroblast
CHROMOSOME 22													
GM5878		R1640	15I	0148	28	M	WH 46,XY,t(10;22)(10pter>10q26.3::22q11.2>22qter;22pter>22q11.2::10q26.3>10qter)	B	A	PARENT			Clinically normal; father of a child with DiGeorge Syndrome; proband not in Repository
GM6275		R1640	15I	0148	29	F	WH 46,XX,t(11;22)(11pter>11q23::22q11>22qter;22pter>22q11::11q23>11qter)	B	A	PROBAND			One child has der(22) syndrome
GM2324		R1640	20I	0182	27	F	WH 46,XX,t(16;22)(16qter>16p13::22q12>22qter;22pter>22q21::16p13>16pter),9qh+	B	A	MOTHER	179	2	Mother of GM2325 Fibroblast
*GM3197		R1640	20I	0318	31	F	WH 46,XX,inv(5)(pter>p13::q13>p13::q13>qter),t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND	518	1	No hematological disorder; habitual aborter; normal phenotype; see GM3196 Fibroblast
GM6228		R1640	15I	0148	9MO	F	47,XX,+der(22),t(11;22)(22pter>22q11::11q23.3>11qter)mat	U	A	PROBAND	933	1	Hypotonia, developmental delay, ear pits, long philtrum, microretrognathia, high palate, long tapered fingers, slight frontal bossing, and small upslanted palpebral fissures; mother is GM6229 Lymphoid

CHROMOSOMALLY ABERRANT HUMAN LYMPHOBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							TRANSLOCATION						
CHROMOSOME 22													
GM6229		R1640	15I	0148	23	F	WH 46,XX,t(11;22)(11pter>11q23.3::22q11> 22qter;22pter>22q11::11q23.3>11qter)	B	A	MOTHER	933	2	Clinically normal; balanced carrier
GM6943		R1640	15I	0074	24	F	WH 46,XX,t(21;22)(21qter>21p12::22q11.1> 22qter;22pter>22q11.1::21p12>21pter)	B	A	PARENT	977	1	Son has unbalanced translocation and Down's Syndrome; see GM6944 Fibroblast
							RECOMBINANT						
CHROMOSOME 11													
*GM5913A		R1640	15I	0294	3	M	WH 46,XY,rec(11)(pter>q22::p14>p11.2::q22> qter)mat	U	A	SIB	889	2	Macular dysfunction, cleft lip and palate, and developmental delay; has triplication of 11p11.2>p14; 157% of normal RBC Catalase activity; mother is GM5912A Lymphoid
CHROMOSOME 13													
*GM5750		R1640	15I	0294	7	F	WH 46,XX,rec(12),dir ins(12;13)(12pter> 12p11.2::13q12.3>13q22::12p11.2>12qter) mat	U	A	SIB	863	1	Mental retardation; trisomic for 13q12.3 >13q22; normal RBC values for LDH, TPI, & PepB; normal electrophoresis patterns for LDH, PepB, TPI, and ESD; no Retinoblastoma; mother is GM5751 Lymphoid
							MARKER CHROMOSOME						
CHROMOSOME X													
GM5854		R1640	15I	0148	60	F	WH 46,X,fra(X)(q27) 4% of cells show fra(X)	B	A	GRMOTHER	819	4	Carrier for X-linked mental retardation; one affected son; two affected grandsons; 1% of lymphocytes and 4% of lymphoid cells show fra(X); proband is GM5131 Fibroblast
GM5855		R1640	15I	0148	24	F	WH 46,X,fra(X)(q27) 6% of cells show fra(X)	B	A	MOTHER	819	5	Carrier for X-linked mental retardation; two affected sons; one affected brother; mild speech difficulties; proband is GM5131 Fibroblast
CHROMOSOME 10													
*GM5523A		R1640	15I	0418	28	F	WH 46,XX,fra(10)(q25) fra(10)(q25) in 16% of cells with FudR	B	A	PROBAND	855	1	See GM5425A Fibroblast; short stature, late menarche, irregular menses, small uterus, small ovaries, aortic insufficiency, myopia, and cataracts

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMD	FAMM	REMARKS
							MARKER CHROMOSOME						
CHROMOSOME 14													
*GM4761A		R1640	15I	0310	58	M	WH 46,XY,14p+	U	A	PROBAND			Clinically normal; 10% of cells show random chromosome loss; p+ appears to be a homogeneously staining region; HSR contains amplified genes for ribosomal RNA
CHROMOSOME 15													
GM3384		R1640	20I	0077	9	M	WH 49,XXXXY,15q+	U	A	PROBAND			Arab
							INVERSION						
CHROMOSOME X													
GM6626		R1640	15I	0148		F	BL 46,X,inv(X)(pter>p11.4::q22.1>p11.4::q22.1>qter)	B	A	PROBAND			Phenotypically normal; two sons have same inv(X); 30% of cells have additional X chromosome; in all cells the inv(X) was late labelling
CHROMOSOME 05													
*GM3197		R1640	20I	0318	31	F	WH 46,XX,inv(5)(pter>p13::q13>p13::q13>qter),t(17;22)(17qter>17p13::22q11>22qter;22pter>22q11::17p13>17pter)	B	A	PROBAND	518	1	No hematological disorder; habitual aborter; normal phenotype; see GM3196 Fibroblast
CHROMOSOME 09													
*GM1921A		R1640	15I	0061	23	M	WH 47,XY,+21,inv(9)(pter>p11::q13>p11::q13>qter)mat,t(8;14)(8pter>8q11::14q11>14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	See GM1920 Fibroblast; Down's Syndrome phenotype
CHROMOSOME 15													
*GM4347		R1640	15I	0369	38	F	WH 47,XX,+inv dup(15)(pter>q1::q1>pter)	U	A	PROBAND	713	1	Phenotypically normal
							INSERTION						
CHROMOSOME 11													
*GM5912A		R1640	15I	0294	20	F	WH 46,XX,inv ins(11)(pter>p14::p11.2>q22::p14>p11.2::q22>qter)	B	A	PARENT	889	1	Balanced carrier; a daughter had aniridia, multiple anomalies, failure to thrive, and severe psychomotor retardation; son is GM5913A Lymphoid
CHROMOSOME 12													
*GM575Q		R1640	15I	0294	7	F	WH 46,XX,rec(12),dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter) mat	U	A	SIB	863	1	Mental retardation; trisomic for 13q12.3>13q22; normal RBC values for LDH, TPI, & PepB; normal electrophoresis patterns for LDH, PepB, TPI, and ESD; no Retinoblastoma; mother is GM5751 Lymphoid

CHROMOSOMALLY ABERRANT HUMAN LYMPHOBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							INSERTION						
CHROMOSOME 12													
*GM5751		R1640	15I	0294	29	F	WH 46,XX,dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter;13pter>13q12.3::13q22>13qter)	B	A	PARENT	863	2	Phenotypically normal; a son had bilateral Retinoblastoma and multiple anomalies
CHROMOSOME 13													
*GM5750		R1640	15I	0294	7	F	WH 46,XX,rec(12),dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter) mat	U	A	SIB	863	1	Mental retardation; trisomic for 13q12.3>13q22; normal RBC values for LDH, TPI, & PepB; normal electrophoresis patterns for LDH, PepB, TPI, and ESD; no Retinoblastoma; mother is GM5751 Lymphoid
*GM5751		R1640	15I	0294	29	F	WH 46,XX,dir ins(12;13)(12pter>12p11.2::13q12.3>13q22::12p11.2>12qter;13pter>13q12.3::13q22>13qter)	B	A	PARENT	863	2	Phenotypically normal; a son had bilateral Retinoblastoma and multiple anomalies
							ISOCHROMOSOME						
CHROMOSOME X													
GM3543		R1640	20I	0107	15	F	WH 45,X/46,X,i(Xq)	U	A	PROBAND			Turner Syndrome; 40%/60%
							DUPLICATION						
CHROMOSOME 10													
GM5047		R1640	15I	0354	23	M	WH 46,XY,dup(10)(pter>q23.2::q11.2>qter)	U	A	PROBAND	825	1	Spastic diplegia, mental retardation, seizures, and minimal pectus carinatum
CHROMOSOME 15													
*GM4347		R1640	15I	0369	38	F	WH 47,XX,+inv dup(15)(pter>q1::q1>pter)	U	A	PROBAND	713	1	Phenotypically normal
							DICENTRIC CHROMOSOME						
CHROMOSOME Y													
GM6967		R1640	15I	0204	4MO	A	WH 45,X/46,X,dic(Y)(p11)/47,X,dic(Y)dic(Y)	U	A	PROBAND	937	1	Ambiguous genitalia; 45%/37%/16%
CHROMOSOME 15													
GM6246		R1640	15I	0418	5	F	WH 47,XX,+dic(15)(pter>q13::q13>pter)	U	A	PROBAND			Developmental delay, small, abnormal EEG, subnormal intelligence, and microcephaly
							DERIVATIVE CHROMOSOME						
CHROMOSOME 01													
GM6226		R1640	15I	0148	6	M	WH 46,XY,-1,+der(1),t(1;16)(1pter>1q44::16p13.11>16pter)mat	U	A	PROBAND	932	1	Hb alpha/Hb beta ratio equals 3:2 in RBC

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER REL TO PROBAND	FAM#	FAMM#	REMARKS
							DERIVATIVE CHROMOSOME					
CHROMOSOME 02												
GM7158		R1640	15I	0491	2	M	OT 46,XY,-2,+der(2),t(1;2)(2pter>2q37::1q32.1>1qter)mat	U	A PROBAND	876	1	Severely retarded and dysmorphic; Fumarate hydratase activity consistent with trisomy for 1q
CHROMOSOME 09												
*GM5518		R1640	15I	0294	31MO	F	WH 46,XX,-9,+der(9)(9qter>9p22::11p13>11pter),del(11)(qter>p11.1::)	U	A PROBAND			Aniridia-Wilms tumor; mental retardation, failure to thrive, sporadic; no family history for Aniridia-Wilms; 50% of RBC Catalase act; norm LDH-A act; norm fibroblast Hb epsilon DNA genes; app mono for 11p11.1>11p13 & 9pter>9p22
*GM1388		R1640	20I	0061	14	F	WH 46,XX,-13,+der(9),t(9;13)(9pter>9q22::13q12>13qter)mat or 46,XX,-13,+der(9),t(9;13)(9pter>9q21::13q12.1>13qter)mat	U	A PROBAND	369	1	Profoundly retarded; clinical features of trisomy 9p syndrome; see GM1387 Fibroblast
CHROMOSOME 12												
*GM0892A		R1640	20I	0024	12	F	WH 45,XX,-18,der(12),t(12;18)(12qter>12p11::18q11>18qter)/47,XX,+12	U	A PROBAND			HLA type A2(A9),B12,B18; clinically normal; 22% of cells are 47,XX,+12
GM2820		R1640	20I	0107	7MO	F	BL 46,XX,-12,+der(12),t(9;12)(12pter>12q24::9p13>9pter)mat	U	A PROBAND	324	1	See GM2819 Fibroblast
CHROMOSOME 21												
GM6136		R1640	15I	0148	2	M	WH 46,XY,-21,+der(21),t(10;21)(21pter>21q22.3::10p11.2>10pter)mat	U	A PROBAND	908	2	Multiple congenital anomalies; bilateral cleft lip and palate; absent patellae; severe colconeo valgus and metatarsus abductus; mother is GM6135 Lymphoid
CHROMOSOME 22												
GM6228		R1640	15I	0148	9MO	F	47,XX,+der(22),t(11;22)(22pter>22q11::11q23.3>11qter)mat	U	A PROBAND	933	1	Hypotonia, developmental delay, ear pits, long philtrum, microretrognathia, high palate, long tapered fingers, slight frontal bossing, and small upslanted palpebral fissures; mother is GM6229 Lymphoid
							DELETION					
CHROMOSOME X												
*GM7773		R1640	15I	0039	28	F	WH 46,X,del(X)(pter>21.3::21.1>qter)	U	A PROBAND			Mild mental retardation; normal gonadal development; patient has partial OTC deficiency and heterozygosity for Chronic Granulomatous disease; lymphocytes & fibroblasts show random X inactivation; in lymph cult, del(X) is late replicating

CHROMOSOMALLY ABERRANT HUMAN LYMPHOBLASTS

DELETION

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
CHROMOSOME 01													
*GM0605A		R1640	15I	0025	24	F	WH 46,X,+15,del(1)(1pter>1p32::1p22>1qter)/47,XX,+15,del(1)(1pter>1p32::1p22>1qter)	U	A	PROBAND			32% of cells are missing an X chromosome; culture established from an apparently normal individual; abberations probably arose in vitro
CHROMOSOME 06													
GM6802		R1640	15I	0148	2MO	M	WH 46,XY,del(6)(pter>q25:)	U	A	PROBAND			Craniofacial dysmorphia; both parents have normal karyotypes
CHROMOSOME 09													
GM5067		R1640	15I	0326	3	M	BL 47,XY,+del(9)(9pter>9q11:)mat	U	A	PROBAND	602	1	Multiple malformations and mental retardation; see GM3226 Fibroblast
GM5066		R1640	15I	0326	34	F	BL 46,XX/47,XX,+del(9)(9pter>9q11:)	U	A	MOTHER	602	2	Short stature; mentally slow; 42%/58%; see GM3225 Fibroblast
CHROMOSOME 10													
GM3470		R1640	20I	0120	1	F	WH 46,XX,del(10)(pter>p13::p12>qter)	U	A	PROBAND	239	2	Microcephaly, microphthalmia, and hypotelorism; father is GM3469 Lymphoid; both parents have normal karyotypes
GM6936		R1640	15I	0418	27MO	F	WH 46,XX,del(10)(qter>p13:)	U	A	PROBAND	974	1	Developmental delay and dysmorphic features; parents both have normal karyotypes; see GM6937 Fibroblast
CHROMOSOME 11													
*GM5518		R1640	15I	0294	31MO	F	WH 46,XX,-9,+der(9)(9qter>9p22::11p13>11pter),del(11)(qter>p11.1:)	U	A	PROBAND			Aniridia-Wilms tumor; mental retardation, failure to thrive, sporadic; no family history for Aniridia-Wilms; 50% of RBC Catalase act; norm LDH-A act; norm fibroblast Hb epsilon DNA genes; app mono for 11p11.1>11p13 & 9pter>9p22
*GM6803		R1640	15I	0107	18	M	WH 46,XY,del(11)(pter>p13::p11.2>qter)	U	A	PROBAND			Aniridia, mental retardation, hypogonadism, abnormal right kidney, high arched palate, microphallus, cryptorchidism, & dysplastic testes; 50% of normal mother's Catalase activity
*GM3316		R1640	20I	0311	3	F	WH 46,X,del(11)(p11.2p1300),t(X;11)(Xpter>Xq11.1::11p1300>11pter;11qter>11p11.2::Xq11.1>Xqter)	U	A	PROBAND	617	1	South American; normal X is late replicating; mental retardation with bizarre dysmorphology syndrome; see GM2859 Fibroblast

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	RAL UNBAL	VER	REL TO PROBAND	FAM#	FAMM#	REMARKS
							DELETION						
CHROMOSOME 11													
GM7736		R1640	15I	0148	1	M	WH 46,XY,del(11)(pter>p13::p14.1>qter)	U	A	PROBAND	1074	1	AGR triad; micropenis; cryptorchidism; bilateral aniridia; optic nerve atrophy; see GM6938B Fibroblast
CHROMOSOME 13													
GM3887		R1640	15I	0016	1	F	WH 46,XX,del(13)(pter>q22:)	U	A	PROBAND			13q- syndrome
GM7890		R1640	15I	0312	3	F	WH 46,XX,del(13)(pter>q12.3::q21.2>qter)	U	A	PROBAND			Developmental delay; growth retardation; mild facial dysmorphism; normal eye exam; 50% of normal Esterase D activity in blood
GM1484		R1640	20I	0094	27MO	F	WH 46,XX,del(13)(pter>q14.1::q22.1>qter)	U	A	PROBAND	187	1	Bilateral Retinoblastoma; sporadic; see GM1142A Fibroblast & GM3022 SV40 Transformed Fibroblast
GM7312		R1640	15I	0094	4	F	46,XX,del(13)(pter>q14.1::q21.2>qter)	U	A	PROBAND	1025	1	Bilateral Retinoblastoma; sporadic; negative family history; normal growth and development except for delayed speech; see GM5877 Fibroblast
CHROMOSOME 18													
GM2054		R1640	20I	0194	3	M	WH 46,XY,del(18)(pter>q21:)	U	A	PROBAND			
CHROMOSOME 20													
*GM7945		R1640	10U	0455	3MO	M	WH 46,XY,del(20)(pter>q13.1:)	U	A	PROBAND			Multiple malformations; seizures; failure to thrive; hemizygous range of Adenosine Deaminase enzyme activity in RBC, fibroblasts, and lymphoblast culture
							ANEUPLOID						
CHROMOSOME X													
*GM0605A		R1640	15I	0025	24	F	WH 46,X,+15,del(1)(1pter>1p32::1p22>1qter)/47,XX,+15,del(1)(1pter>1p32::1p22>1qter)	U	A	PROBAND			32% of cells are missing an X chromosome; culture established from an apparently normal individual; abberations probably arose in vitro
*GM1202B		R1640	15I	0066	7	M	WH 49,XXXXY	U	A	PROBAND			
GM3384		R1640	20I	0077	9	M	WH 49,XXXXY,15q+	U	A	PROBAND			Arab

CHROMOSOMALLY ABERRANT HUMAN LYMPHOBLASTS

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM#	REMARKS
							ANEUPLOID						
CHROMOSOME X													
GM3543		R1640	20I	0107	15	F	WH 45,X/46,X,i(Xq)	U	A	PROBAND			Turner Syndrome; 40%/60%
*GM1416B		R1640	15I	0061	27	F	WH 48,XXXX	U	A	PROBAND	370	1	Develop delay; mod retard; delayed language develop; abnormal dermatoglyphics; bilat epicanth folds; sm mandible; bilat dermato-lipomas; lens opacities; high arched palate; mildly hypotonic; normal menses; see GM1415E Fibroblast
GM6061A		R1640	15I	0078	25MO	F	WH 49,XXXXX	U	A	PROBAND	914	1	Developmental delay; microcephaly; small for age; low set ears; bilateral epicanthal folds; decreased muscle tone; low total finger-ridge count; parents both have normal karyotypes; see GM5882 Fibroblast
CHROMOSOME 09													
GM5067		R1640	15I	0326	3	M	BL 47,XY,+del(9)(9pter>9q11:)mat	U	A	PROBAND	602	1	Multiple malformations and mental retardation; see GM3226 Fibroblast
GM5066		R1640	15I	0326	34	F	BL 46,XX/47,XX,+del(9)(9pter>9q11:)	U	A	MOTHER	602	2	Short stature; mentally slow; 42%/58%; see GM3225 Fibroblast
CHROMOSOME 10													
GM1566		R1640	20I	0008	16	F	WH 45,XX,-10/46,XX,-10,+12/47,XX,+12	U		PROBAND	406	1	Cystathioninuria; HLA type A1,A28,B13, Bw35; 4%/30%/66%; parents are GM1456A & GM1868 Lymphoid
CHROMOSOME 12													
*GM0892A		R1640	20I	0024	12	F	WH 45,XX,-18,der(12),t(12;18)(12qter>12p11::18q11>18qter)/47,XX,+12	U	A	PROBAND			HLA type A2(A9),B12,B18; clinically normal; 22% of cells are 47,XX,+12
GM1990		R1640	20I	0089	51	F	WH 46,XX/47,XX,+12	U	A	PROBAND	359	1	Clinically normal; 64% of cells are trisomic for chromosome 12; formerly GM1077
GM1566		R1640	20I	0008	16	F	WH 45,XX,-10/46,XX,-10,+12/47,XX,+12	U		PROBAND	406	1	Cystathioninuria; HLA type A1,A28,B13, Bw35; 4%/30%/66%; parents are GM1456A & GM1868 Lymphoid

REP #	PAS #	CULTURE MEDIA	SMTR CODE	AGE	SEX	RACE	PARIS NOMENCLATURE	BAL UNBAL	VER	REL TO PROBAND	FAMS	FAMM	REMARKS
							ANEUPLOID						
CHROMOSOME 15													
*GM0605A	R1640	15I	0025	24	F	WH	46,X,+15,del(1)(1pter>1p32::1p22>1qter)/47,XX,+15,del(1)(1pter>1p32::1p22>1qter)	U	A	PROBAND			32% of cells are missing an X chromosome; culture established from an apparently normal individual; abberations probably arose in vitro
CHROMOSOME 21													
GM1201A	R1640	15I	0144	14MO	F	WH	45,XX,-21	U	A	PROBAND			46,XX,-21,+t(21;21) in peripheral blood of a Down's Syndrome patient; 4% random chromosome loss
GM3716	R1640	15I	0343	23	M	WH	47,XY,+21	U	A	PROBAND			Down's Syndrome
*GM1921A	R1640	15I	0061	23	M	WH	47,XY,+21,inv(9)(pter>p11::q13>p11::q13>qter)mat,t(8;14)(8pter>8q11::14q11>14qter;14pter>14q11::8q11>8qter)	U	A	PROBAND	418	1	See GM1920 Fibroblast; Down's Syndrome phenotype
GM4927B	R1640	15I	0061	27	M	WH	47,XY,+21	U	A	PROBAND	781	1	Down's Syndrome phenotype; 47,XY,+21,fra(16)(q22) in leukocytes; no fra(16) observed in lymphoblastoid culture; see GM4928 Fibroblast
CHROMOSOME 22													
*GM2184A	R1640	15I	0186	36	M	WH	46,XY/45,XY,-22	U	A	PROBAND	455	1	Clinically normal spouse of a Huntington's disease patient; 1 cell out of 50 showed 47,XY,+18; 50% of cells are 46,XY; see GM2185 Fibroblast

Human Amniotic Fluid Cell Cultures

Human Amniotic Fluid Cell Cultures

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAMS	FAMNS	REMARKS
APPARENTLY NORMAL HUMAN AMNIOTIC FLUID CELL CULTURES												
Apparently Normal (Amniotic)												
*GM0472	9	R1640 30I	0058	19FW	M		++	A	PROBAND			46+XY
*GM0473	6	R1640 30I	0058	17FW	M		++	A	PROBAND			46+XY; 8% of cells show random chromosome loss
GM0474	10	R1640 30I	0058	18FW	M		++	A	PROBAND			46+XY
*GM0956	4	R1640 20U	0064	17FW	F		++	A	PROBAND			46+XX
*GM0957A	6	EEMEM 20U	0064	18FW	F		++	A	PROBAND			46+XX; 10% of cells show random chromosome loss
BIOCHEMICALLY MUTANT HUMAN AMNIOTIC FLUID CELL CULTURES												
Cystinosis: Early Onset (Nephropathic Type; Infantile Type) - 21980												
GM0804		EEMEM 20U	0121	22FW	M	WH	--	B	PROBAND			46+XY; increased intracellular level of 1/2 cystine; passage 3 at IMR
Fabry Disease (Diffuse Angiokeratoma; Hereditary Dystopic Lipidosis) - 30150												
GM0636	8	HEMEM 20U	0017	18FW	M	WH	Y-	B	PROBAND			46+XY; Alpha-galactosidase deficient
Galactosemia (Transferase Deficiency) - 23040												
GM1743	3	EEMEM 20U	0077	24FW	M	WH	--	B	PROBAND	32	1	46+XY; see GM1741 Fibroblast
Homocystinuria (Cystathionine Synthetase Deficiency) - 23620												
GM0919	6	EEMEM 20U	0038	30FW	F		--	B	PROBAND			46+XX; mother is also affected; deficient cystathionine synthetase activity
Hypophosphatasia (Phosphoethanolaminuria) - 24150												
GM3336		EEMEM 20U	0321	20FW	F	WH	--	A	PROBAND	604	1	46+XX; severe in utero onset form; deficient alkaline phosphatase; passage 5 at IMR; parents are GM3327 & GM3328 Fibroblasts; see also GM3502 Fibroblast
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800												
*GM2338	5	EEMEM 20U	0183	15FW	M	WH	Y-	A	PROBAND	74	1	46+XY; see GM2290 and GM2291 Fibroblast and GM2292A Lymphoid; two similarly affected male sibs and a maternal uncle
Fragile X-linked Mental Retardation: Martin - Bell Syndrome - 30955												
GM5132	4	EEMEM 20U	0148	FW	F	WH	+-		SIS	819	2	Mother is an obligate carrier for X-linked mental retardation; brother is GM5131 Fibroblast; two brothers and a maternal uncle have X-linked mental retardation; 6% of peripheral blood leukocytes show fra(X)
Metachromatic Leukodystrophy: Infantile (Arylsulfatase A Deficiency) - 25010												
GM2095		EEMEM 20U	0105	18FW	F	AI	--		PROBAND			Passage 3 at IMR; after delivery showed increased urinary sulfatides

HUMAN AMNIOTIC FLUID CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMN#	REMARKS
BIOCHEMICALLY MUTANT HUMAN AMNIOTIC FLUID CELL CULTURES												
Metachromatic Leukodystrophy: Infantile (Arylsulfatase A Deficiency) - 25010												
GM2214		EEMEM 20U	0105	20FW	M	WH	??		PROBAND			At risk; passage 4 at IMR; sister has typical infantile MLD
Mucolipidosis: Type IV (Ganglioside Sialidase Deficiency) - 25265												
*GM2529	19	EEMEM 20U	0077	19FW	F	WH	--	B	AFF SIS	229	2	Ashkenazi; abnormal cytoplasmic lysosomal storage bodies; affected sib is GM2048 Fibroblast and GM2533 Lymphoid; 46,XX; 8% of cells show random chromosome loss
Mucopolysaccharidosis: Type II - Hunter Syndrome - 30990												
GM1584	6	EEMEM 20U	0077	21FW	M	WH	Y-	A	AFF BRO	247	2	46,XY; deficient iduronate sulfatase; Israeli; affected brother is GM1583 Fibroblast
CHROMOSOMALLY ABERRANT HUMAN AMNIOTIC FLUID CELL CULTURES												
Aneuploid Chromosome Number - Non-Trisomic												
GM1993	7	EEMEM 20U	0172	21FW	M	WH		A	PROBAND			47,XYY
GM2269	9	EEMEM 20U	0109	18FW	M	WH		A	PROBAND			47,XXY
GM2587	5	EEMEM 20U	0172	17FW	M	WH		A	PROBAND			47,XYY
GM3535	5	EEMEM 20U	0172	20FW	M	WH		A	PROBAND			47,XXY
GM3091	4	EEMEM 20U	0172	15FW	M	WH		A	PROBAND	448	1	47,XXY; see GM3102 Fibroblast
Chromosome Deletion												
GM2421A	8	EEMEM 20U	0172	16FW	F	WH		A	PROBAND			46,XX,del(13)(qter>p11::)mat; unbalanced; newborn was phenotypically normal; mother is also clinically normal
GM6312		EEMEM 20U	0418	33FW	M	WH		A	PROBAND			Abnormal ultrasound at 29-37 weeks gestation; polyhydramnios; brachycephaly; abdominal obstruction; 46,XY,del(13)(pter>q21.2::); passage 1 at IMR; child was born with multiple congenital anomalies
Inverted Chromosome												
GM4536	5	EEMEM 20U	0224	15FW	F	OT		A	PROBAND			46,XX,inv(2)(pter>p21::q21>p21::q21>qter),inv(9)(pter>p11::q13>p11::q13>qter); balanced; Black/Caucasian; phenotypically normal
GM2055	7	EEMEM 20U	0172	17FW	F			A	PROBAND			Balanced; 46,XX,inv(9)(pter>p11::q13>p11::q13>qter); father shows same inv(9) in blood; family history of spontaneous abortions and infertility

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
CHROMOSOMALLY ABERRANT HUMAN AMNIOTIC FLUID CELL CULTURES												
Inverted Chromosome												
GM3367	7	EEMEM 30I	0172	16FW	F	OR		A	PROBAND			Balanced; 46,XX,inv(18)(pter>p11::q21> p11::q21>qter)pat
GM2380	6	EEMEM 20U	0273	18FW	F			A	SIS	481	2	Balanced; 46,XX,inv(13)(pter>p11::q22> p11::q22>qter)pat; sister with unbalanced aberration is GM1570 Fibroblast
Marker Chromosome												
GM2662	6	EEMEM 20U	0172	21FW	F	BL		A	PROBAND			47,XX,+mar; unbalanced; marker is de novo and satellited
GM3090		EEMEM 20U	0172	23FW	F	WH		A	PROBAND			Bilateral simian crease; incurving fifth finger; dislocated hip; 46,XX,21q+; passage 2 at IMR
Recombinant Chromosome												
*GM4627	5	EEMEM 20U	0003	20FW	M	WH		A	AFF BRO	560	2	Mother has inv(3)(p25q21); 46,XY,rec(3),dup q,inv(3)(qter>p25::q21>qter),9qh+; unbalanced; brother is GM0366 Fibroblast
Translocated Chromosome												
GM0477	9	EEMEM 20U	0048	14FW	F	WH		A	PROBAND			46,XY,-18,+der(18),t(13;18)(18pter>18q 213 or q23::13q12 or q14>13qter); unbalanced; phenotypically female
GM3435		EEMEM 20U	0273	15FW	M			A	PROBAND			46,XY,-4,+der(4),t(4;11)(4qter>4p14::11 p15>11pter)mat; unbalanced; passage 3 at IMR
GM5564	4	EEMEM 20U	0224	15FW	F	WH		A	PROBAND			Both parents have normal karyotypes; 46,XX,t(3;9)(3pter>3q29::9p22>9pter;9qter>9p22::3q29>3qter); balanced
GM6321	5	EEMEM 20U	0326	17FW	M	WH		A	PROBAND			46,XY,t(9;22)(9pter>9q13::22q13.3>22qter;22pter>22q13.3::9q13>9qter); balanced
GM3100	8	EEMEM 20U	0172	18FW	F	WH		A	PROBAND	449	1	46,XX,t(9;17)(9qter>9p22::17p11>17pter;17qter>17p11::9p22>9pter)mat; balanced; mother is GM3119 Fibroblast
Aneuploid Chromosome Number - Trisomy												
GM2419	7	EEMEM 20U	0172	16FW	F	WH		A	PROBAND			47,XX,+21
GM2422	4	EEMEM 20U	0172	16FW	F	WH		A	PROBAND			47,XX,+18
GM3250	7	EEMEM 20U	0172	15FW	M	WH		A	PROBAND			47,XY,+18

HUMAN AMNIOTIC FLUID CELL CULTURES

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	AGE	SEX	RACE	GENETIC STATUS	VERIFIED	RELATION TO PROBAND	FAM#	FAMM#	REMARKS
CHROMOSOMALLY ABERRANT HUMAN AMNIOTIC FLUID CELL CULTURES												
Aneuploid Chromosome Number - Trisomy												
GM3416	7	EEMEM 20U	0172	16FW	F	OR		A	PROBAND			47,XXX
GM3606	4	EEMEM 20U	0172	18FW	F	OT		A	PROBAND			Black/Caucasian; 47,XX,+21; less sensitive to interferon A and B compared to normal diploid fibroblasts
GM3673	7	EEMEM 20U	0342	19FW	F	BL		A	PROBAND			47,XX,+del(9)(pter>q21:)mat
GM3699	5	EEMEM 20U	0172	18FW	F	IN		A	PROBAND			47,XXX
GM3769	5	EEMEM 20U	0172	16FW	M	WH		A	PROBAND			47,XY,+18

Animal Cell Cultures, Hybridomas, and Somatic Cell Hybrids

Animal Cell Cultures, Hybridomas, and Somatic Cell Hybrids

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Bovine Tissue							
GM3655	2	EEMEM 20U	0336				Skin fibroblast culture established from a 5 year old apparently normal Holstein cow; 60,XX in fibroblasts
GM5864	8	EEMEM 20I	0428				Fibroblast culture from a 2 yr old male Limousin with bovine protoporphyria; photosensitivity and high levels of protoporphyrin in erythrocytes and feces
*GM5865	8	EEMEM 20I	0428				Fibroblast culture from a 5 year old female 7/8 Limousin 1/8 Hereford with bovine protoporphyria; photosensitivity & high levels of protoporphyrin in erythrocytes & feces
GM5868B	5	EEMEM 20U	0428				Fibroblast culture from a 2 year old female Holstein with congenital erythropoietic porphyria; photosensitivity and high levels of protoporphyrin in erythrocytes and feces
GM5867	7	EEMEM 20I	0428				Fibroblast culture from a 3 year old female Holstein; clinically normal carrier for congenital erythropoietic porphyria
*GM5868	8	EEMEM 20I	0428				Fibroblast culture from a 6 year old female 1/2 Limousin 1/2 Hereford; clinically normal carrier for bovine protoporphyria
GM6034	3	EEMEM 20U	0428				Fibroblast culture from a clinically normal 3 year old male Hereford
GM6035	4	EEMEM 20U	0428				Fibroblast culture from a clinically normal 2 year old female Holstein
Bovine Endothelial Cell Cultures							
*GM3905		EEMEM 20U	0163		1034	1	Clinically normal; male; 6 mos. gestation; aortic endothelial cell culture; received at PDL 53 and passaged 3 more times at IMR; estimated replicative life-span is 80 PDL; clone BFA-1c
*GM7372		EEMEM 10U	0163		1034	2	Clinically normal; male; 6 mos. gestation; aortic endothelial cell culture transformed in vitro with benzo(a)pyrene; BFA-1C 1BPT flat transformants; submitted at PDL 283; non-tumorigenic in nude mice

ANIMAL CELL CULTURES, HYBRIDOMAS, AND SOMATIC CELL HYBRIDS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Bovine Endothelial Cell Cultures							
*GM7373		EEMEM 10U	0163		1034	3	Clinically normal; male; 6 mos. gestation; aortic endothelial cell culture transformed in vitro with benzo(a)pyrene; SFA-1C 1BPT multilayered transformants; submitted at PDL 355; tumorigenic in nude mice
Feline Tissue							
*GM6206	3	EEMEM 20U	0336				Skin fibroblast culture from a 6 yr old male domestic short-haired cat with a dominantly inherited syndrome of fragility & hyperextensibility of skin resembling Ehlers-Danlos syndrome with abnormal collagen packing in fibrils & fibers
GM6207	2	EEMEM 20U	0336				Clinically normal two year old male cat; fibroblast culture; 38,XY
Canine Tissue							
GM3978	2	EEMEM 20U	0325				Clinically normal adult, female mongrel; fibroblast culture
GM3983	3	EEMEM 20U	0325				Five year old male Cocker Spaniel; clinically normal; fibroblast culture
Chinese Hamster							
*GM0215		EEMEM 05U	0022				Line V79; young adult male, estab. by Ford and Yerganian; cloned by Elkind; fibroblast culture established from a lung biopsy; near diploid
*GM0458		EEMEM 10U	0053		965	1	Line CHW; male; fibroblast cell line established by spontaneous transformation of a normal skin culture; pseudodiploid; 30% pseudotetraploid; 22,Y
*GM0459	2	EEMEM 10U	0053	A	965	1	Line CHW-1102, MMS induced mutant of GM0458, selected with 8-azaguanine, HGPRT deficient; fibroblast culture
Syrian Baby Hamster Kidney							
*GM0345		EEMEM 10U	0083	B			Line TG-2, HGPRT deficient, selectable with 6-thioguanine, heteroploid, fibroblast culture
*GM0348A		EEMEM 10U	0083	B	873	1	Line B-1; fibroblast culture; thymidine kinase deficient; selectable with 5-bromodeoxyuridine; heteroploid

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Syrian Baby Hamster Kidney							
*GM0511		EEMEM 10U	0083	B	873	1	Line A-5, subclone of B-1 (GM0348A); increased folate reductase; thymidine kinase deficient; fibroblast culture; heteroploid
Mouse Tissue							
*GM0086D		EEMEM 15U	0040	A			Friend erythroleukemic cell line, clone #745; DMSO inducible; derived from DBA/2 mouse; grows in suspension
*GM0346A		EEMEM 20U	0083	A			Line A-9; mouse L cell; fibroblast-like; HGPRT & APRT deficient; selectable with 8-azaguanine; heteroploid
*GM0347A		EEMEM 20U	0083	A			Line B-82, mouse L cell; fibroblast-like; thymidine kinase deficient; selectable with 5-bromodeoxyuridine; heteroploid
*GM0979		HF12 10I	0158	A			Murine erythroleukemic line, T3-C12, derived from a splenic focus in DDD Mouse infected with Friend Leukemia virus; suspension culture; inducible for erythroid differentiation by DMSO; aneuploid; hypotetraploid
*GM5267		EEMEM 10U	0096	A			SV40 transformed tfm mouse kidney cells; HPRT deficient; selectable with 6-thioguanine; expresses testicular feminization locus mutation - little detectable androgen binding activity; modal number of 38 chromosomes
*GM5373		R1640 15I	0413	B			Suspension culture derived from the SEWA tumor which was induced by polyoma virus in an A.SW mouse; originally an osteosarcoma; modal no. of 43 chromosomes; XY; 34% of cells are polyploid; 50% of cells show double minute chromosomes
*GM5384	48	EEMEM 20U	0415	B			Y-1; epithelial-like cell culture established from an adrenal cortex tumor of a male LAF1 mouse; secretes steroid hormones; reverse transcriptase positive; 40,XY,1q+; 40% of cells have 1 or 2 centric fragments
*GM5862	35	EEMEM 20U	0051	B			Mouse line 3T6; fibroblast cell line established by Todaro and Green from Swiss albino mouse embryos; secretes collagen and hyaluronic acid

ANIMAL CELL CULTURES, HYBRIDOMAS, AND SOMATIC CELL HYBRIDS

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Ovine Tissue							
GM3550	2	EEMEM 20U	0325				Southdown sheep; female; 4 months old; fibroblast culture
GM4053	2	EEMEM 20U	0350				Clinically normal, adult, pregnant sheep; fibroblast culture
Nonhuman Primates							
GM3442	3	EEMEM 20U	0325		535	1	Stumptail; old world female monkey; fibroblast culture from skin biopsy; same monkey as GM3443; 42,XX in fibroblasts
GM3443		R1640 20I	0325		535	1	Stumptail; old world female monkey; lymphoblast culture; same monkey as GM3442; 42,XX in lymphoblast culture
GM3444	2	EEMEM 20U	0325		536	1	Stumptail; old world male monkey; fibroblast culture from skin biopsy
GM3446	2	EEMEM 20U	0325		537	1	Crab-eating Macaque; old world male monkey; fibroblast culture from skin biopsy; 42,XY
GM3448	3	EEMEM 20U	0325		538	1	Chimpanzee; old world monkey; 7 yr old male; fibroblast culture from skin biopsy; 48,XY
GM3450	4	EEMEM 20U	0325		539	1	Chimpanzee; old world monkey; 15 yr old male; fibroblast culture from skin biopsy; 48,XY
GM3452	3	EEMEM 20U	0325		540	1	Chimpanzee; old world male monkey; fibroblast culture from skin biopsy; 48,XY
GM4272	3	EEMEM 20U	0220				Sumatran Urangutan; 10 yr old male; skin fibroblast culture; 48,XY in leukocytes and fibroblasts; homozygous for Sumatran chromosome #2 polymorphism
GM6213	1	EEMEM 20U	0220		928	1	Clinically normal; 26 year old male orangutan; skin fibroblast culture; 48, XY; homozygous for Sumatran chromosome #2 polymorphism in blood leukocytes and fibroblasts
GM7404		R1640 15I	0051				Marmoset cell line; grows as a semi-suspension culture; infected and shedding B95-8 virus; cell free supernatant may be used to transform human B-lymphocytes to produce immortal lymphoid cell cultures

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Myeloma (Plasmacytoma) Cell Lines for Hybridomas							
*GM3569B		DUMEM 10U	0189	B	853	1	Mouse myeloma, line SP2/0; HPRT deficient; non-immunoglobulin producing; medium contains 4.5g glucose per liter
*GM3570C		DUMEM 10U	0189	B			Mouse myeloma; line P3X63Ag8653; HPRT deficient; non-immunoglobulin producing; derived from P3 by Rajewsky; medium contains 4.5g glucose per liter
*GM3571		DUMEM 10U	0189	B			BALB/C mouse myeloma; line P3X63Ag8; secretes IgG1(K) (MOPC21); HPRT deficient; original Milstein line; medium contains 4.5g glucose per liter
*GM3572		DUMEM 10U	0189	B			Mouse myeloma; line 45.6TG1.7; HPRT deficient; produces IgG2(K); derived by Scharff; medium contains 4.5g glucose per liter
*GM3573A		DUMEM 10U	0189	B			BALB/C mouse myeloma, line P3X63NS1; HPRT deficient, produces Kappa, non-immunoglobulin secreting; medium contains 4.5g glucose per liter
GM5426A		R1640 10U	0419	B	853	1	Clone of mouse myeloma line SP2/0 (GM3569B) selected for good growth in 8-azaguanine; HPRT deficient
*GM5657		DUMEM 10U	0189	B			Mouse myeloma; line S194; derived from an IgA (alpha, kappa) secreting myeloma which arose after mineral oil induction in a Balb/c mouse; synthesizes heavy and light chain but no immunoglobulin is secreted; resistant to bromodeoxyuridine
Hybridomas							
►*GM3566		DUMEM 20U	0339	A			Lewis rat spleen cells fused with mouse myeloma P3X63Ag8; line D4.37; produces rat IgM,"non-Kappa"; mouse IgGp, Kappa; antigen specificity: RT1-N.A; medium contains 4.5g glucose per liter
►*GM3568		DUMEM 20U	0339	A			Lewis rat spleen cells fused with mouse myeloma P3X63Ag8; line D4.69; produces rat IgG2c, Kappa & mouse IgG1, Kappa; antigen specificity: RT1-N.A; medium contains 4.5g glucose per liter
►*GM3704		DUMEM 10I	0339	A			Lewis rat spleen cells fused with mouse myeloma SP2/0; line PnK2.13.2; antigen specificity: RT1-N.A; medium contains 4.5g glucose per liter & gamma globulin free horse serum instead of fetal bovine serum

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Hybridomas							
►*GM3705		DUMEM 10I	0339	A			Lewis rat spleen cells fused with mouse myeloma P3X63Ag8; antigen specificity: RT1-N.A; line PhN2.11.6; medium contains 4.5g glucose per liter & gamma globulin free horse serum instead of fetal bovine serum
►*GM4278		DUMEM 10U	0189	B			C57B16 mouse immunized with cultured human neuroblastoma cells fused with mouse myeloma P3X63Ag8; produces antibody (IgM) against human neuroblastoma and B and null cell leukemias; line PI153/3; antibody binds complement
►*GM4279		DUMEM 10U	0189	B			C57B16 mouse immunized with human B-cell line supernatant fused with mouse myeloma P3X63Ag8; produces cytotoxic antibody (IgM) against human Ia; line P3B13C2; medium contains 4.5g glucose per liter
► GM4280		DUMEM 10U	0189	B			Balb/C mouse immunized with human B-cell line fused with mouse myeloma P3X63Ag8; produces cytotoxic antibody (IgG2a) against human HLA:A,B,& C; line KE2; medium contains 4.5g glucose per liter
Human - Chinese Hamster Somatic Cell Hybrid							
► GM6317		EEMEM 20U	0053	A			Human male fetal fibroblast X HPRT deficient Chinese Hamster (CHW1103); originally selected in THAG medium plus ouabain; only intact human chromosome is Y; contained genes for human PGM2 (CHR#4), KAR (CHR#12), & NP (CHR#14) at early pas
► GM6318A		EEMEM 20U	0053	A			Human fetal fibroblast X HPRT deficient Chinese Hamster (CH65691) using Sendai virus; selected in HAT medium; contains only the human X chromosome and no other human isozyme markers; culture grows as a monolayer
► GM7297	4	EEMEM 20U	0006	A			Human X HPRT deficient Chinese Hamster lung cell line; hybrid # 11.1.2.2; selected in medium containing azaserine; contains only human chromosomes # 3 & X; at P6, 98% of cells have human X & 74% have human # 3 chromosome
► GM7298	4	EEMEM 20U	0006	A			Human X HPRT deficient Chinese Hamster lung cell line; hybrid # 2.12; selected in medium containing azaserine; at passage 6, contains human chromosomes # 4 (26%), 14 (64%), 20 (12%), 21 (60%), & t(Xili)(Xqter>cen>1iqter) (96%)

REP #	PASSAGE #	CULTURE MEDIA	SUBMITTER CODE	VERIFIED	FAM#	FAMM#	REMARKS
Human - Chinese Hamster Somatic Cell Hybrid							
► GM7299	5	EEMEM 20U	0006	A			Human X HPRT deficient Chinese Hamster lung cell line; hybrid # MR 8.2; selected in medium containing azaserine; contains human chromosomes # 1 & X; at P7, X is present in 15/22 cells, 2/22 cells have # 1 & X, & 1/22 cells has X and # 6
► GM7300	4	EEMEM 20U	0006	A			Human X HPRT deficient Chinese Hamster lung cell line; hybrid # 1.11; selected in medium containing azaserine; contains human chromosomes # 6, 8, 11, & X; 94%, 85%, 94%, & 90% respectively at P6
► GM7301	4	EEMEM 20U	0006	A			Human X HPRT deficient Chinese Hamster lung cell line; hybrid # 16.2; selected in medium containing azaserine; at passage 7, 48% of cells have human X and 38% have human # 12 chromosomes; original hybrid also contained the human Y
Human - Rat Somatic Cell Hybrid							
► GM6853A		R1640 20U	0039	A			Human fetal liver fibroblast X Rat nepatoma line 7777-14b-AZA; selected in HAT medium; retains the human X chromosome; negative for GPI, ADA, & ACO2; culture grows in suspension

Statement to Users of Somatic Cell Hybrids

Recipients of somatic cell hybrids are cautioned that the cell cultures are not warranted to be free of unrecognized human DNA sequences and may also be unstable. Characterization of these lines is performed at a single passage after recovery of frozen stock from liquid nitrogen and this may not apply at subsequent passages. Neither the Repository nor the original submitter can be responsible for changes that may occur when the hybrids are grown and utilized in other laboratories. Stability may particularly be affected by conditions during shipping and by the use of other than the specified culture media.

Recipients are urged to re-characterize the hybrids in their own laboratories and to share their results with the Repository. A data base of information received on hybrid characterization will be maintained by the Repository with open access to all interested researchers.

Recipients are also asked to cite the Repository as the source of the hybrid as well as the Repository identification number for the hybrid in any publications resulting from the use of the hybrid and to forward reprints to the Repository.

Recipients may not obtain hybrid cells for commercial purposes without the written approval of the original submitter and the NIGMS project officer.

NIA Aging Cell Repository

NIA
Aging Cell
Repository

NIA Aging Cell Repository

The potential value of cultured mammalian cells as a model system in aging research was first emphasized in the early 1960's following the demonstration that human fibroblast-like cells display a finite growth potential *in vitro*. Subsequently, an inverse relationship was found between growth potential of skin fibroblast cultures and chronological donor age. Later observations documenting the diminished growth potential of cultures derived from individuals with some genetic disorders that mimic premature aging (segmental progeroid syndromes) further reinforced the perception of the utility of *in vitro* cell culture systems in biogerontologic research. In addition, it has been emphasized that a variety of genetic disorders display at least some features of accelerated aging. These developments dramatically increased the demand for cultured cells from a variety of genotypes, some of which are very rare and therefore difficult to obtain. In response to the increasing demand from the scientific community, the National Institute of Child Health and Human Development, which at that time had responsibility for research on aging, awarded a contract in 1974 to the Institute for Medical Research to establish a cell repository. The contract was renewed in 1976 under the sponsorship of the newly created National Institute on Aging, and again in 1980.

This present catalog for the Aging Cell Culture Repository lists many categories of cell cultures for cellular aging studies. Paramount in the listings are the specially characterized diploid human fetal lung fibroblast lines. In this category are IMR-90 (female) and IMR-91 (male) which were developed expressly for the NIA Cell Culture Repository and are available at low, intermediate and high population doubling levels. Sufficient early passage cells of these two lines are preserved so that they should be available to the research community for many years. Also in this category is WI-38 (female) which was the first diploid cell strain developed and utilized for the study of aging at the cellular level and human virus vaccine production. WI-38 is available at population doubling levels 14, 22, 34 and 44. The Repository also distributes TIG-1 (female) which was developed in Japan for use in aging research. MRC-5 (male) from Britain, another well characterized diploid human fetal lung fibroblast cell strain, is available at three population doubling levels. All of the above cultures have been widely utilized for aging studies, and an extensive literature details the role they have played in research on the causes and effects of aging at the cellular level as well as many other biomedical studies. Other diploid human fetal fibroblast cultures, including matched skin and lung cultures from the same individuals, are listed in the catalog.

Many normal human non-fetal fibroblast cultures are available

at early passage from the Repository. Included in this group are skin fibroblasts derived from participants in the Baltimore Longitudinal Study of the Gerontology Research Center (GRC). These cultures were derived from healthy donors, with biopsies taken from the same anatomical site, and initiated using similar, standardized procedures (Cytogenet. Cell Genet. 31:40, 1981). The in vitro lifespan for each GRC culture has been estimated (Proc. Natl. Acad. Sci. USA 75:1353, 1978) and the population doubling level of the stored cultures is available. The ages of the donors range from 17 to 96 years, with numerous donors in their 7th and 8th decades of life. Many of these cultures have been karyotyped. The GRC cultures have and will continue to provide an important resource for testing the validity of the diploid fibroblast as a model for cellular aging.

Because of the importance of "segmental progeroid syndromes" including Werner, Hutchinson-Guilford, Cockayne and Rothmund-Thomson syndromes for aging research, the Repository has made the banking and distribution of these rare genetic diseases a high priority. Cultures from individuals with these diseases provide valuable resource material which the majority of interested investigators would find difficult to obtain on their own. During the past two years the Repository added a number of nonhuman primate skin fibroblast cultures which should provide useful material for aging studies because the close phylogenetic relationship of the donors to humans, the wide range of lifespans available in the donor species and the well-defined phasing out of cellular multiplication that has been observed in nonhuman primate cultures. The ten cultures from young and old rhesus monkeys should provide especially valuable material for cellular aging studies. This year the Repository has added bovine and equine endothelial, smooth muscle and fibroblast cultures and canine and porcine endothelial cell cultures to its listings. Additional cultures of these types will be added during the coming year.

Instructions for ordering cell cultures are given on page X. Recipients of grants or contracts from the National Institute on Aging may order IMR-90, IMR-91 and cell cultures prefixed by AG at no charge, if the NIA grant or contract number is given when orders are placed. Orders from all other individuals will be charged the fee stated on page X.

The NIA Aging Cell Repository is supported by contract AG-0-2100.

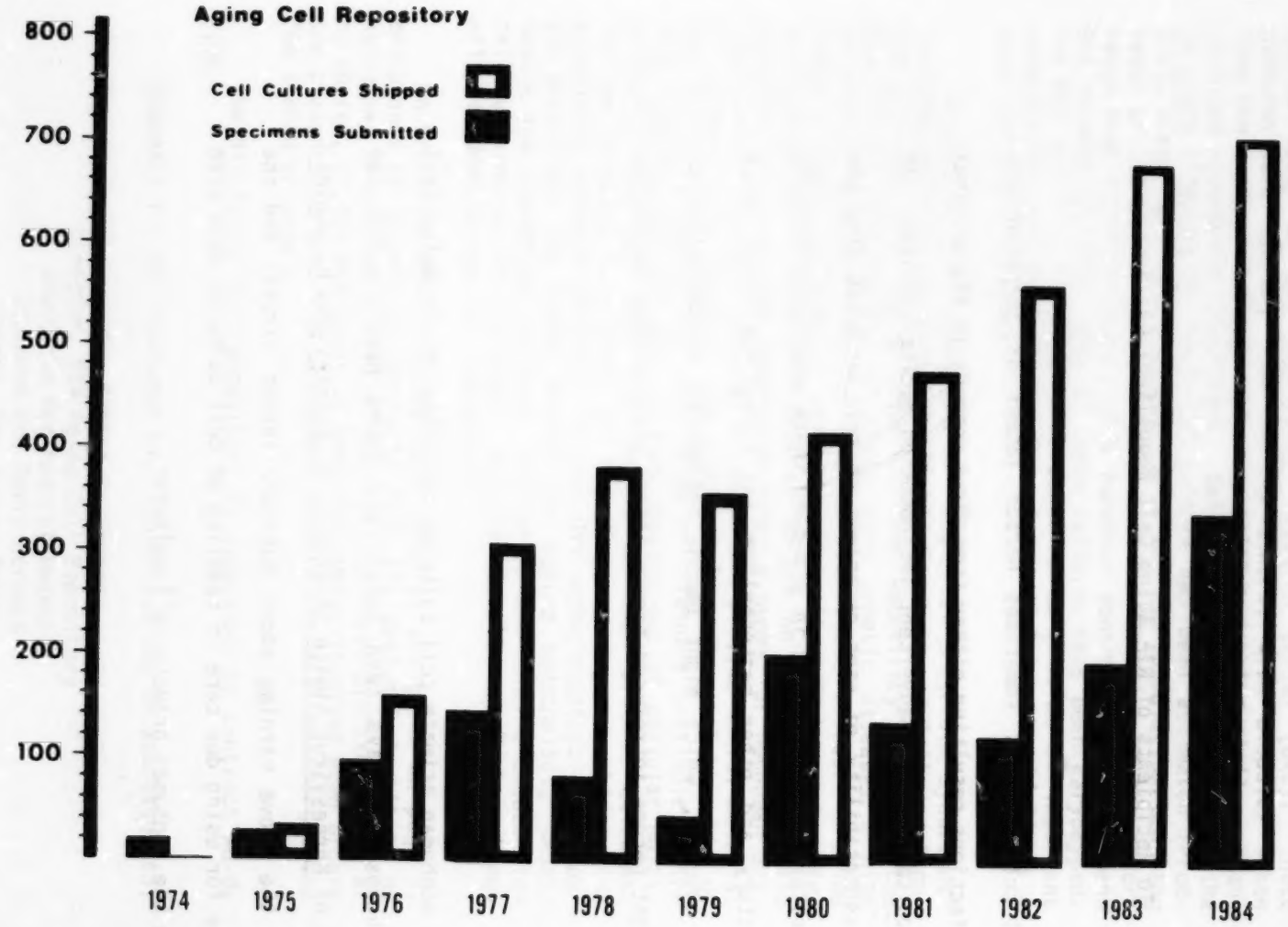
Requests for cell cultures and information may be addressed to:

Dr. Arthur E. Greene
Director, NIA Aging Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103
Telephone 609-966-7377

To Recipients of NIA Aging Cell Repository Cells

Cell cultures can sometimes harbor latent or inappropriate virus infections resulting either from virus genomes in the original tissue source or from inadvertent introduction during handling. It is the responsibility of each recipient of cell cultures from the NIA Repository to handle them in a manner which minimizes exposure of laboratory and other personnel to any infectious agent which might be present, which might become introduced accidentally, or which might become introduced from a latent state within the cells.

All nonhuman primate cell cultures available in the Aging Cell Culture Repository have been tested and found negative for the presence of Herpesvirus simiae (B-virus), H. saimiri and H. ateles. However, the above warning about possible latent viruses and the directive for using due care in handling of cell cultures must also apply to the nonhuman primate cell cultures.


Aging Cell Repository
Cell Cultures Shipped
Specimens Submitted
800
700
600
500
400
300
200
100
1974
1975
1976
1977
1978
1979
1980
1981
1982
1983
1984

Explanation of the Data Columns for the Aging Cell Culture Repository Sections

COLUMN

REP#: The Repository number of the cell culture is the number assigned to a culture upon receipt at or upon initiation by the Aging Cell Culture Repository. It consists of two letters and four digits, i.e., AG1234. The exceptions are IMR-90 and IMR-91 cultures for which the Rep# is I90 or I91 followed by P and two digits. These last two digits are the passage number for the culture. Thus, I90 P05 is IMR-90 frozen at passage 5. When it becomes necessary to expand the supply of an original cell line, the expansion is designated by adding a letter to the Repository number, i.e., AG1234A. The expansion may differ from the original cell line in that its passage number may be higher.

AGE: The age of the individual donor at the time the biopsy or blood specimen is taken. It is displayed as years months days. For a fetal tissue-derived culture, an estimated gestational age is displayed.

SEX: Phenotypic sex; Male (M), Female (F), or Ambiguous (A).

RACE: WH-Caucasian; BL-Black; OR-Oriental: IN-Indian (India); AI-American Indian; BR-Polynesian. Space is left empty if race is unknown.

CELL TYPE: The assignment of cell type is based on a culture's cell morphology and tissue of origin. If a culture has been further characterized for differentiated cell functions, these characterizations are described in the remarks section.

ORIGIN: Either the biopsy site, organ, or tissue of origin for the material used to initiate the culture.

PDL FROZEN: The number of cumulative population doublings accrued by the culture from primary culture to freeze pool population. Generally, cultures initiated from explants are designated PDL=0 for cells of the primary culture at confluency. For cultures initiated by enzymatic dissociation, the cells plated or the cells that attach in the primary flask are designated PDL=0.

IN VITRO LIFE-SPAN: The total number of cumulative population doublings a culture can be expected to achieve before reaching senescence and phase-out of cell division. This number includes the population doublings accumulated prior to freezing. In some cases this determination has been obtained by actually subculturing to the end of the in vitro life-span of the culture, and

in other cases the determination has been estimated by colony size distribution assays. In vitro life-span determinations are affected by media, serum, split-ratios and other culturing variables. The space is left blank if nc life-span determinations have been made.

MEDIA: Culture medium. For explanation of the code see Appendix A, page 469. This column describes the recommended growth medium and percentage of fetal bovine serum.

GENETIC STATUS:

+	Normal gene
-	Mutant gene
Y-	Hemizygous for X-linked trait
+?	Genetic status not determined
+ -(0)	Carrier for trait as determined by pedigree or clinical diagnosis

The McKusick* number immediately following the name of the disease indicates the dominant, recessive, or X-linked nature of the disease. The first digit is one for a dominant disease (e.g. Alzheimer Disease - 10430), two for a recessive disease (e.g. Werner Syndrome - 27770), and three for an X-linked disease (e.g. Dyskeratosis Congenita - 30500).

RELATION TO PROBAND
OR REL TO PROBAND: Example: AFF BRO = Affected brother.

FAM#: A unique pedigree number is assigned if cultures from more than one member of a family or more than one sample type (i.e. fibroblast, lymphoblast, endothelial, etc.) from a single individual are stored in the Repository.

FAMM#: Within each pedigree, each family member is assigned a unique family member number. Cultures having the same FAMM# were obtained from the same individual.

REMARKS: Pertinent information on donor, biopsy and culture.

SUBMITTER CODE: A number which identifies the investigator who submitted the specimen or in some cases the culture to the Repository. For code and submitter names with addresses see Appendix B, page 470.

Special Notations

* Before a Repository number indicates that a literature reference for this cell line is found in Appendix C, page 490.

AGING CELL REPOSITORY CULTURES

SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES

REP#	CELL CULTURE DESIGNATION	PDL FROZEN	SEX	AGE YR MO DA	RACE	MEDIA
*I90 P05	IMR-90 - Normal Human Fetal Lung Fibroblast	12	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152. Earlier passage IMR-90 cells available only with permission of NIA: see guidelines in Science 196:60-63 (1977).

*I90 P07	IMR-90 - Normal Human Fetal Lung Fibroblast	21	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*I90 P08	IMR-90 - Normal Human Fetal Lung Fibroblast	24	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*I90 P09	IMR-90 - Normal Human Fetal Lung Fibroblast	27	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*I90 P10	IMR-90 - Normal Human Fetal Lung Fibroblast	30	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

AGING CELL REPOSITORY CULTURES

SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES

REPO	CELL CULTURE DESIGNATION	PDL FROZEN	SEX	AGE YR MO DA	RACE	MEDIA
*I90 P11	IMR-90 - Normal Human Fetal Lung Fibroblast	33	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*I90 P13	IMR-90 - Normal Human Fetal Lung Fibroblast	37	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*I90 P15	IMR-90 - Normal Human Fetal Lung Fibroblast	41	F	00 04 00	WH	MC5A 20U

The IMR-90 line was established on 7/7/75 using explants of minced lung tissue obtained from an apparently normal 16 week fetus. The karyotype is 46,XX; normal diploid female. No variant isoenzyme patterns found for lactate dehydrogenase, malate dehydrogenase, 6-phosphogluconate dehydrogenase, and glucose-6-phosphate dehydrogenase; G6PD is isoenzyme type B. The HLA typing is A2, B12, BW40, B5. Reverse transcriptase was not detected. The maximum number of population doublings achieved to senescence range from 58-73. Plating efficiency is 36% at PDL 10, 48% at PDL 15 & 0.5% at PDL 45. Culture initiated and submitted by #0152.

*AG28048	IMR90-SV-40 Virus-Transformed Fetal Lung Fibroblast	108	F	00 04 00	WH	EEMEM 20U

AG28048 is a post-crisis SV40 transformed IMR90 culture. It was derived from the same pool of infected cells that initiated the pre-crisis transformed IMR90 culture, AG3204. Both cultures were initiated by infecting IMR90 at PDL 30 with SV40 virus strain Rh 911. AG28048 contains 100% T-antigen positive cells & 1% to 5% V-antigen positive cells. The cell morphology is epithelial-like. Culture submitted by #0135.

AG3204	IMR90-SV-40 Virus-Transformed Fetal Lung Fibroblast	38	F	00 04 00	WH	EEMEM 20U

AG3204 is a pre-crisis SV40 transformed IMR90 culture. It was derived from the same pool of infected cells that initiated the post-crisis transformed IMR90, AG28048. Both cultures were initiated by infecting IMR90 at PDL 30 with SV40 virus strain Rh 911. AG3204 contains 10% T-antigen positive cells & a low frequency of V-antigen positive cells. The morphology of the cells is a mixture of fibroblast-like and epithelial-like. Culture submitted by #0135.

*I91 P05	IMR-91 - Normal Human Fetal Lung Fibroblast	15	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

AGING CELL REPOSITORY CULTURES

SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES

REP#	CELL CULTURE DESIGNATION	PDL FROZEN	SEX	AGE YR MO DA	RACE	MEDIA
*I91 P07	IMR-91 - Normal Human Fetal Lung Fibroblast	19	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

*I91 P08	IMR-91 - Normal Human Fetal Lung Fibroblast	22	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

*I91 P09	IMR-91 - Normal Human Fetal Lung Fibroblast	26	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

*I91 P10	IMR-91 - Normal Human Fetal Lung Fibroblast	30	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

*I91 P11	IMR-91 - Normal Human Fetal Lung Fibroblast	32	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to 76. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

*I91 P13	IMR-91 - Normal Human Fetal Lung Fibroblast	37	M	00 03 00	WH	MC5A 20U

The IMR-91 lung-derived line was initiated on 2/24/76 from explants of minced tissue of both lungs from an approximately 12 week fetus. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The reported number of population doublings achieved to senescence range from 49 to .6. The HLA typing is A2, A11, B18 and B5. G6PD is isoenzyme type B and 6PGD is isoenzyme type A. Culture submitted by #0152.

AGING CELL REPOSITORY CULTURES

SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES

REP#	CELL CULTURE DESIGNATION	PDL FROZEN	SEX	AGE YR MO DA	RACE	MEDIA
*AG5965B	MRC-5 - Normal Human Fetal Lung Fibroblast	15	M	00 03 15	WH	EEMEM 20U

The MRC-5 cell line was developed in September 1966 from lung tissue taken from a 14 week fetus removed for psychiatric reason from a 27 year old physically healthy woman. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Cumulative population doublings to senescence is 42-48. G6PD isoenzyme is type B. Isolated and submitted by #0445.

*AG5965C	MRC-5 - Normal Human Fetal Lung Fibroblast	27	M	00 03 15	WH	EEMEM 20U

The MRC-5 cell line was developed in September 1966 from lung tissue taken from a 14 week fetus removed for psychiatric reason from a 27 year old physically healthy woman. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Cumulative population doublings to senescence is 42-48. G6PD isoenzyme is type B. Isolated and submitted by #0445.

*AG5965D	MRC-5 - Normal Human Fetal Lung Fibroblast	40	M	00 03 15	WH	EEMEM 20U

The MRC-5 cell line was developed in September 1966 from lung tissue taken from a 14 week fetus removed for psychiatric reason from a 27 year old physically healthy woman. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Cumulative population doublings to senescence is 42-48. G6PD isoenzyme is type B. Isolated and submitted by #0445.

*AG6173A	TIG-1 - Normal Human Fetal Lung Fibroblast	17	F	00 05 00	OR	EEMEM 20U

The TIG-1 cell line was initiated at the Tokyo Metropolitan Institute of Gerontology on 7/28/76 using lung tissue from a 5 month gestational age fetus from a therapeutic abortion. Cells released by a trypsin digestion of a lung specimen were used for the primary culture. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. The maximum number of population doublings achieved to senescence range from 55 to 79. G6PD is isoenzyme type B. Submitted by #0444.

*AG6814D	WI-38 - Normal Human Fetal Lung Fibroblast	14	F	00 03 00	WH	EEMEM 20U

The WI-38 cell line was developed in July 1962 by L. Hayflick from lung tissue taken from a therapeutically aborted fetus of about 3 months gestational age. Cells released by a trypsin digestion of the lung tissue were used for the primary culture. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A maximum life-span of 50 population doublings for this culture was obtained at this repository. A thymidine labelling index of 95% was obtained after recovery. G6PD is isoenzyme type B. This culture of WI-38 is an expansion from frozen passage 9 cells obtained from submitter #0460.

*AG6814G	WI-38 - Normal Human Fetal Lung Fibroblast	22	F	00 03 00	WH	EEMEM 20U

The WI-38 cell line was developed in July 1962 by L. Hayflick from lung tissue taken from a therapeutically aborted fetus of about 3 months gestational age. Cells released by a trypsin digestion of the lung tissue were used for the primary culture. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A maximum life-span of 50 population doublings for this culture was obtained at this repository. A thymidine labelling index of 95% was obtained after recovery. G6PD is isoenzyme type B. This culture of WI-38 is an expansion from frozen passage 9 cells obtained from submitter #0460.

AGING CELL REPOSITORY CULTURES

SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES

REP#	CELL CULTURE DESIGNATION	PDL FROZEN	SEX	AGE YR MO DA	RACE	MEDIA
*AG6814H	WI-38 - Normal Human Fetal Lung Fibroblast	34	F	00 03 00	WH	EEMEM 20U

The WI-38 cell line was developed in July 1962 by L. Hayflick from lung tissue taken from a therapeutically aborted fetus of about 3 months gestational age. Cells released by a trypsin digestion of the lung tissue were used for the primary culture. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A maximum life-span of 50 population doublings for this culture was obtained at this repository. A thymidine labelling index of 86% was obtained after recovery. G6PD is isoenzyme type B. This culture of WI-38 is an expansion from frozen passage 9 cells obtained from submitter #0460.

*AG6814I	WI-38 - Normal Human Fetal Lung Fibroblast	44	F	00 03 00	WH	EEMEM 20U

The WI-38 cell line was developed in July 1962 by L. Hayflick from lung tissue taken from a therapeutically aborted fetus of about 3 months gestational age. Cells released by a trypsin digestion of the lung tissue were used for the primary culture. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A maximum life-span of 50 population doublings for this culture was obtained at this repository. A thymidine labelling index of 64% was obtained after recovery. G6PD is isoenzyme type B. This culture of WI-38 is an expansion from frozen passage 9 cells obtained from submitter #0460.

AG7217	WI-38-SV-40 Virus-Transformed Fetal Lung Fibroblast		F	00 03 00	WH	EEMEM 15U

Line WI-38 VA13, subline 2RA is a post-crisis SV40 virus-transformed derivative of WI-38, & was derived 3/64 by infecting WI-38 cells at passage 23 with SV40 virus. Loss of contact inhibition occured 48 days post-infection and crisis occurred 20 weeks post-infection. T-antigen positive cells are present, and the karyotype is hyperdiploid. G6PD isoenzyme is type B. The culture is frozen at passage 255, and was submitted by #0507.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7720	24 00 00	F	GRC#737		9	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7721	25 00 00	F	GRC#738		6	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7124	26 00 00	F	GRC#695		10	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XX/47,XX,+8/48,XX,+8,+20; 79%/12%/10%.

AG7306	28 00 00	F	GRC#698		10	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7719	28 00 00	F	GRC#735		9	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7715	31 00 00	F	GRC#723		8	26	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5244	32 00 00	F	GRC#432		8	45	PROBAND	877	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,22p+; diploid female. The donor is the daughter of AG4153 and the sister of AG4438.

AG7471	32 00 00	F	GRC#710		8	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REPO	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7472	33 00 00	F	GRC#711		5	47	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7718	33 00 00	F	GRC#733		7	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7121	34 00 00	F	GRC#686		9	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XX,9qh+/46,XX,9qh+, del(10)(qter>p13:); 84%/16%.

AG5838A	36 00 00	F	GRC#575		7	54	GR DAU	899	3	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,22p+. Donor is the daughter of AG5807A and the granddaughter of AG4064.

AG7308	36 00 00	F	GRC#700		10	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7716	39 00 00	F	GRC#727		7	31	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5245	40 00 00	F	GRC#435		14	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.

AG7307	40 00 00	F	GRC#699		8	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7311	42 00 00	F	GRC#706		7	26	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG5836A	44 00 00	F	GRC#522		8	43	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG5837A	47 00 00	F	GRC#571		10	39	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG7118	47 00 00	F	GRC#652		11	33	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG7304	48 00 00	F	GRC#689		9	24	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
*AG5270	50 00 00	F	GRC#448		9	47	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG5839A	50 00 00	F	GRC#530		12	47	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG5840A	50 00 00	F	GRC#537		12	42	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7473	50 00 00	F	GRC#712		7	45	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG7302	52 00 00	F	GRC#683		10	39	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG5275	53 00 00	F	GRC#446		9	42	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG7139	53 00 00	F	GRC#665		8	34	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG7305	53 00 00	F	GRC#690		10	39	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG7807	53 00 00	F	GRC#743		8	28	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG7714	56 00 00	F	GRC#720		7	37	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG5844A	57 00 00	F	GRC#584		12		PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,21s+.									

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7140	57 00 00	F	GRC#666		9	32	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7477	58 00 00	F	GRC#717		7	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7310	60 00 00	F	GRC#702		8	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5265	61 00 00	F	GRC#438		9	31	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,21s+ with 50% of examined cells showing random chromosome abnormalities.

AG6010	62 00 00	F	GRC#541		8	46	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.

AG5246	65 00 00	F	GRC#444		8	33	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XX/46,XX,t(11;19)(11pter>11q13::19q13>19qter;19pter>19q13::11q13>11qter), balanced; 62%/38%.

AG5266	65 00 00	F	GRC#439		8	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,15s+.

AG7141	66 00 00	F	GRC#675		9	28	PROBAND			EEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG5813B	67 00 00	F	GRC#496		9	23	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX,9qh+.

AG7119	68 00 00	F	GRC#673		9	23	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.

AG5806B	70 00 00	F	GRC#506		15	51	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.

AG7309	70 00 00	F	GRC#701		9	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7479	70 00 00	F	GRC#719		8	49	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5811B	71 00 00	F	GRC#605		11	53	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.

AG7120	72 00 00	F	GRC#676		9	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX;1qh-,16qh+.

AG6952	73 00 00	F	GRC#606		9	53	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7122	73 00 00	F	GRC#687		10	25	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
AG7303	74 00 00	F	GRC#688		7	34	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG7474	84 00 00	F	GRC#714		7	33	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG5247C	87 00 00	F	GRC#445		9	58	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XX; diploid female.									
*AG6234	17 00 00	M	GRC#440		9	40	SON	939	2	HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. The donor is the son of AG6235.									
AG7804	23 00 00	M	GRC#736		10	32	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
AG6310	27 00 00	M	GRC#568		8	38	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.									
*AG4054	29 00 00	M	GRC#173-1		9	45	PROBAND			HEMEM 20U
	The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.									

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4441A	29 00 00	M	GRC#260-2		8	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG5415A	29 00 00	M	GRC#288		18	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7478	29 00 00	M	GRC#718		7	35	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG4151	30 00 00	M	GRC#220-1		9		PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(2;18)(2pter>2q33::18q21>18qter;18pter>18q21::2q33>2qter); 60%/40%, balanced.

*AG4062	31 00 00	M	GRC#210-2		8	40	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG5412	31 00 00	M	GRC#306		10	17	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY,1qh+; diploid male.

*AG4150	32 00 00	M	GRC#213-1		13	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY with 10% of cells showing inv(1).

AG6285	32 00 00	M	GRC#531		7	51	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6962	32 00 00	M	GRC#604		8		PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The donor is the son of AG5807A, the grandson of AG4064 and a brother of AG5838A.

*AG4438	33 00 00	M	GRC#257-1		13	44	BRO	877	2	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. The donor is the son of AG4153 and the brother of AG5244.

AG5411	33 00 00	M	GRC#305		11	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6954	33 00 00	M	GRC#609		7	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4557	34 00 00	M	GRC#255-1		8	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY,inv(1)(q11q21)/46,XY,inv(1)(q11q21),t(1;13)(1pter>1q25::13q12>13qter;13pter>13q12::1q25>qter), balanced; 96%/4%.

*AG4447A	35 00 00	M	GRC#247-1		9	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4553	38 00 00	M	GRC#250-1		8	33	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY with 25% of cells showing random chromosome abnormalities.

AG6278	38 00 00	M	GRC#508		10	55	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6877	38 00 00	M	GRC#569		8	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6880	38 00 00	M	GRC#578		9	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4443	39 00 00	M	GRC#262-3		13	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4656	39 00 00	M	GRC#221-1		9	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Random chromosome abnormalities were observed in 18% of cells examined. Donor is diabetic and hypertensive. He had psoriasis, but was not treated with psoralen-UVA.

AG7476	39 00 00	M	GRC#716		8	16	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG4462A	40 00 00	M	GRC#202-2		10	49	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4354	41 00 00	M	GRC#166-2		10	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4654	41 00 00	M	GRC#205-2		7	43	PROBAND	2007	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(7;12)(q22;q13), balanced/46,XY,t(1;9)(q32;q12), balanced; 76%/16%/8%. A culture initiated from a biopsy taken 5 years later from the same donor is AG7724.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6296	41 00 00	M	GRC#551		8	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4145	42 00 00	M	GRC#183-1		7	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4352	42 00 00	M	GRC#164-1		11	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,t(1;8)(p22;q13), balanced; 96%/4%.

AG5416	42 00 00	M	GRC#293		10	43	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6235	42 00 00	M	GRC#441		7	46	PROBAND	939	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. Donor is the father of AG6234.

AG6307	42 00 00	M	GRC#561		8	47	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4063	43 00 00	M	GRC#211-1		8	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4556	43 00 00	M	GRC#254-1		8	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY,1qh+/46,Y,1qh+,t(X;1)(q24;q25), balanced; 28%/72%.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REPO	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG5189	43 00 00	M	GRC#223-2		10	43	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,2q+; 88%/12%.

*AG4060	44 00 00	M	GRC#158-2		7	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(2;13)(p13;q32), balanced; 88%/12%.

*AG4351	44 00 00	M	GRC#163-1		11	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6282	44 00 00	M	GRC#517		7	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6237	45 00 00	M	GRC#461		9	32	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG7724	46 00 00	M	GRC#768		9	51	PROBAND	2007	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 5 years earlier from the same donor is AG4654.

*AG4440A	47 00 00	M	GRC#259-2		10	28	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY,9qh-.

*AG4446	48 00 00	M	GRC#246-2		11	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6956	48 00 00	M	GRC#612		12	50	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4147	49 00 00	M	GRC#161-1		10	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4456	49 00 00	M	GRC#195-2		11	41	PROBAND	878	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(1;6)(q25;q13), balanced; 44%/56%. Donor is the son of AG4459.

*AG4655	49 00 00	M	GRC#207-1		8	42	PROBAND	840	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG5188	49 00 00	M	GRC#222-1		10	20	PROBAND	901	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 3 years later from the same donor is AG5805B.

AG6295	49 00 00	M	GRC#550		7	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY with 6% of cells being 46,XY,inv(4)(p14q12).

*AG4442	50 00 00	M	GRC#261-3		13	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6878	50 00 00	M	GRC#572		11	53	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7136	51 00 00	M	GRC#447		9	40	PROBAND	1001	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 2 years later from the same donor is AG7144.

*AG4453	52 00 00	M	GRC#192-1		8	39	PROBAND	879	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Donor is the son of AG4065.

AG5103	52 00 00	M	GRC#186-2		7	25	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY,+1qh+.

AG5805B	52 00 00	M	GRC#625		9	40	PROBAND	901	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 3 years earlier from the same donor is AG5188.

AG6306	52 00 00	M	GRC#560		9	47	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4058	53 00 00	M	GRC#176-1		10	29	PROBAND	880	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Donor is the son of AG4144.

*AG4149	53 00 00	M	GRC#212-1		7	35	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4439	53 00 00	M	GRC#256-2		9	37	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG5097	53 00 00	M	GRC#242-2		8	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7144	53 00 00	M	GRC#732		8	29	PROBAND	1001	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 2 years earlier from the same donor is AG5188.

AG5101	54 00 00	M	GRC#187-2		11	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a mosaic with the karyotype 46,XY/46,XY,inv(7)(q22q32),t(9;17)(q34;q21.3) balanced/46,XY,t(9;17)(q34;q21.3) balanced; 52%/32%/16%.

AG5191	54 00 00	M	GRC#217-1		8	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5410	54 00 00	M	GRC#304		8	22	PROBAND	2006	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 4 years later from this same donor is AG7723.

AG6304A	54 00 00	M	GRC#557		9	40	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5192	55 00 00	M	GRC#218-2		11	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6283A	55 00 00	M	GRC#519		10	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6287	55 00 00	M	GRC#535		8	50	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4148	56 00 00	M	GRC#167-1		8	58	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5186	56 00 00	M	GRC#203-2		7	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6305	56 00 00	M	GRC#559		8	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4146	57 00 00	M	GRC#181-1		11	33	PROBAND	999	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 5 years later from this same donor is AG7123.

*AG4385	57 00 00	M	GRC#232-2		10	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4458	57 00 00	M	GRC#197-1		11	28	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7723	58 00 00	M	GRC#750		9	41	PROBAND	2006	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 4 years earlier from the same donor is AG5410.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4353	59 00 00	M	GRC#165-2		13	40	PROBAND	902	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 4 years later from the same donor is AG5804B.

*AG4380A	59 00 00	M	GRC#227-2		8	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,12p+.

*AG4560	59 00 00	M	GRC#185-2		7	46	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6009	59 00 00	M	GRC#505		7	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6239	59 00 00	M	GRC#477		8	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5419	60 00 00	M	GRC#292		12	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6288	60 00 00	M	GRC#538		7	48	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6241	61 00 00	M	GRC#486		8	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY,22s+.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4350	62 00 00	M	GRC#160-2		10	40	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,1q+, 78%/20% and 2% of examined cells showing random abnormalities.

AG6280	62 00 00	M	GRC#513		7	33	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7123	62 00 00	M	GRC#693		10		PROBAND	999	I	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 5 years earlier from this same donor is AG4146.

*AG4444	63 00 00	M	GRC#264-3		15	43	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG4658	63 00 00	M	GRC#237-2		7	21	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XO with 27% of examined cells showing random chromosome abnormalities.

AG5417	63 00 00	M	GRC#279		11	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5804B	63 00 00	M	GRC#621		17	52	PROBAND	902	I	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 4 years earlier from this same donor is AG4353.

AG6881	63 00 00	M	GRC#581		9	35	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4552A	64 00 00	M	GRC#249-2		10	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5102	64 00 00	M	GRC#209-1		7	22	PROBAND	843	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6882	64 00 00	M	GRC#589		9	30	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4061	65 00 00	M	GRC#159-1		9	53	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(8;18)(8pter>8q11::18q11>18qter;18pter>18q11::8q11>8qter), balanced; 80%/20%.

*AG4357	65 00 00	M	GRC#182-1		9	37	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4551	65 00 00	M	GRC#248-2		7	28	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG4659	65 00 00	M	GRC#219-1		9	29	PROBAND	881	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 4 years later from this same donor is AG5283.

AG6294	65 00 00	M	GRC#548		6	49	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4454	66 00 00	M	GRC#193-1		7	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4461	66 00 00	M	GRC#201-2		8	25	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7803	66 00 00	M	GRC#730		6	43	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4355	67 00 00	M	GRC#168-1		11	40	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6281	67 00 00	M	GRC#515		8	37	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6959	67 00 00	M	GRC#633		8	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4559	68 00 00	M	GRC#184-2		7	40	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5098	68 00 00	M	GRC#241-1		11	46	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6957	68 00 00	M	GRC#619		9	50	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7137	68 00 00	M	GRC#387		9	34	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5095	69 00 00	M	GRC#244-1		8	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5099	69 00 00	M	GRC#188-1		7	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5283	69 00 00	M	GRC#556		9	49	PROBAND	881	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 4 years earlier from this same donor is AG4659.

AG6236	69 00 00	M	GRC#453		9	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4445	70 00 00	M	GRC#265-2		8	20	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6284	70 00 00	M	GRC#521		10	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6301	70 00 00	M	GRC#553		7	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7801A	70 00 00	M	GRC#285		11	27	PROBAND	2005	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 3 years later from the same donor is AG5271A.

*AG4558	71 00 00	M	GRC#256-1		9	23	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,inv(5)(pter>q22::q31>q22::q31>qter); 88%/12%.

AG5807A	71 00 00	M	GRC#630		10	46	SON	899	2	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The donor is the son of AG4064 and the father of AG5838A.

AG6242	71 00 00	M	GRC#500		12	33	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6879	71 00 00	M	GRC#577		9	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6885A	71 00 00	M	GRC#598		9	24	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4379	72 00 00	M	GRC#226-1		8	22	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4455	72 00 00	M	GRC#194-1		9	42	PROBAND	859	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. A culture initiated from a biopsy taken 1 year later from this same donor is AG5414.

*AG4056	73 00 00	M	GRC#171-1		9	35	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/47,XY,+18; 88%/12%.

*AG4459	73 00 00	M	GRC#198-1		9	22	FATHER	878	2	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Donor is the father of AG4456.

AG5096	73 00 00	M	GRC#243-2		8	29	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5271A	73 00 00	M	GRC#514		13	42	PROBAND	2005	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 3 years earlier from the same donor is AG7801A.

AG5414	73 00 00	M	GRC#298		8	36	PROBAND	859	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 1 year earlier from this same donor is AG4455.

*AG4142	74 00 00	M	GRC#174-1		6	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4356	74 00 00	M	GRC#208-1		9	26	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6290	74 00 00	M	GRC#542		7	51	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6886	74 00 00	M	GRC#602		8	46	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4153	75 00 00	M	GRC#206-1		6	23	FATHER	877	3	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. Donor is the father of AG4438 and AG5244.

AG5187A	75 00 00	M	GRC#214-1		7	47	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6289	75 00 00	M	GRC#539		7	25	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6302	75 00 00	M	GRC#554		7	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6883	75 00 00	M	GRC#596		10	51	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4144	76 00 00	M	GRC#177-1		12	36	FATHER	880	2	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. Donor is the father of AG4058.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG7722	76 00 00	M	GRC#745		9	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-uoper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4384	77 00 00	M	GRC#231-1		8	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7135	77 00 00	M	GRC#383		11	45	PROBAND	1000	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY,13s+. A culture initiated from a biopsy taken 3 years later from this same donor is AG7143.

AG4554	78 00 00	M	GRC#251-2		8	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4555	78 00 00	M	GRC#253-1		9	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4378	79 00 00	M	GRC#225-2		8	21	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG4660	79 00 00	M	GRC#240-2		7	31	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6953	79 00 00	M	GRC#607		11	36	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4383	80 00 00	M	GRC#230-1		8	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4387	80 00 00	M	GRC#234-1		7	24	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6240	80 00 00	M	GRC#485		8	52	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7143	80 00 00	M	GRC#741		9	25	PROBAND	1000	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 3 years earlier from this same donor is AG7135.

*AG4055	81 00 00	M	GRC#263-2		9	24	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

*AG4057	81 00 00	M	GRC#172-1		9	33	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG4382	81 00 00	M	GRC#229-1		8	47	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG5100	81 00 00	M	GRC#239-2		10	30	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(17;19)(17qter>17p11::19q133>19qter;19pter>19q133::17p11>17pter), balanced; 26%/74%.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
AG6279	81 00 00	M	GRC#510		7	52	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7802	81 00 00	M	GRC#724		7	32	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4152	82 00 00	M	GRC#216-1		7	38	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4381	82 00 00	M	GRC#228-2		7	29	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6243	82 00 00	M	GRC#502		13	37	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6277	82 00 00	M	GRC#507		8	45	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6292	82 00 00	M	GRC#546		8	42	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4386	83 00 00	M	GRC#233-1		12	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	POL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4457	83 00 00	M	GRC#196-1		9	50	PROBAND	838	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 46,XY/46,XY,t(7;20)(7pter>7q11::20q11>20qter;20pter>20q11::7q11>7qter), balanced; 88%/12%. A culture initiated from a biopsy taken 4 years later from this same donor is AG5248A.

AG5413	83 00 00	M	GRC#294		9	41	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4661	84 00 00	M	GRC#238-2		6	39	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5274	84 00 00	M	GRC#540		8	52	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG6276	84 00 00	M	GRC#504		7	23	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG7475	84 00 00	M	GRC#715		7	23	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

*AG4662	87 00 00	M	GRC#235-2		9	27	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG5248A	87 00 00	M	GRC#520		10	44	PROBAND	838	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. The culture is a genetic mosaic: 45,X/46,XY; 66%/33%. A culture initiated from a biopsy taken 4 years earlier is AG4457.

AGING CELL REPOSITORY CULTURES

HUMAN SKIN FIBROBLAST CULTURES FROM THE GRC LONGITUDINAL STUDY

REP#	AGE YR MO DA	SEX	BIOPSY#	RACE	PDL FROZEN	EST. IN VITRO LIFE-SPAN	RELATION TO PROBAND	FAM#	FAMM#	MEDIA
*AG4065	88 00 00	M	GRC#191-1		8	33	FATHER	879	2	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Donor is the father of AG4453.

AG6303A	88 00 00	M	GRC#555		8	19	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like.

AG6308	89 00 00	M	GRC#564		8	47	PROBAND	2008	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 2 years later from the same donor is AG7725.

AG6291	90 00 00	M	GRC#544		6	44	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

AG7725	91 00 00	M	GRC#800		6	43	PROBAND	2008	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 2 years earlier from the same donor is AG6308.

*AG4064	92 00 00	M	GRC#189-1		9	50	PROBAND	899	1	HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male. Donor is the father of AG5807A and is the grandfather of AG5838A.

*AG4059	96 00 00	M	GRC#179-1		10	29	PROBAND			HEMEM 20U

The biopsy site is the mesial aspect of the mid-upper left arm, taken ante-mortem. The primary culture was initiated from explants of a 2 mm-punch biopsy. The cell morphology is fibroblast-like, and the karyotype is 46,XY; diploid male.

DIFFERENTIATED HUMAN CELL CULTURES: AGING

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	PDL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Fetal Tissue							
AG5337	Epithelial, pigmented	Retina of eye		M		18	30	EEMEM 20U
	Retinal pigmented epithelial line RPE-30 was initiated using explants of retinal tissue from a fetus of 13 cm. crown-rump length obtained 10/13/81. The cell morphology is epithelial-like. Culture submitted by #0388.							
AG5338	Epithelial, pigmented	Retina of eye		F		19	30	EEMEM 20U
	Retinal pigmented epithelial line RPE-28 was initiated using explants of retinal tissue from a fetus of 17 cm. crown-rump length obtained 11/9/81. The cell morphology is epithelial-like. Culture submitted by #0388.							
AG6095	Epithelial, pigmented	Retina of eye	00 03 00	M		142		EEMEM 10U
	Permanent line RPE 18 SV2 was obtained by transformation of a fetus-derived retinal pigmented epithelial cell line with SV-40, str. RH 911 virus. This transformed line appears stable without a crisis phase. It is T-antigen positive and does not pigment. The morphology is epithelial-like. Culture submitted by #0388.							
AG6096	Epithelial, pigmented	Retina of eye	00 03 00	M		102		EEMEM 10U
	Permanent line RPE 28 SV4 was obtained by transformation of a fetus-derived retinal pigmented epithelial cell line with SV-40, str. RH 911 virus. This transformed line appears stable without a crisis phase. It is T-antigen positive and does not pigment. The morphology is epithelial-like. Culture submitted by #0388.							
	Apparently Normal: Non-Fetal Tissue							
*AG7086	Mesothelial, peritoneal	Ascites fluid	26 00 00	F		18	48	MES 20U
	Normal human mesothelial cell strain LP-9. This culture was initiated 10/1/83 by directly plating an ascites fluid cell suspension which had been stored frozen since removal from the peritoneal cavity of an ovarian cancer patient on 9/2/81. The culture medium must be supplemented with 20 nanograms/ml EGF and 0.4 micrograms/ml hydrocortisone. The cell morphology is fibroblast-like in medium with supplements, but is epithelial-like in medium without supplements. The karyotype is 46,XX; normal diploid female. Submitted by #0484.							
AG7090A	Mesothelial, peritoneal	Ascites fluid	58 02 09	F		19	50	MES 20U
	Normal human mesothelial cell strain LP-3. This culture was initiated on 10/1/83 by directly plating an ascites fluid cell suspension which had been stored frozen since removal from the peritoneal cavity of an ovarian cancer patient on 9/19/80. The culture medium must be supplemented with 20 nanograms/ml EGF and 0.4 micrograms hydrocortisone. The cell morphology is fibroblast-like in medium with supplements, but is epithelial-like in medium without them. The karyotype is 46,XX; normal diploid female. Culture submitted by #0484.							

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	POL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Fetal Tissue							
AG4450	Fibroblast, lung	Lung	00 03 00	M		13		EEMEM 20U

The culture was initiated on 5/20/80 from explants of minced lung tissue taken from a therapeutically aborted fetus of 12 weeks gestational age. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. A matched skin-derived fibroblast culture from same fetus is AG4449. Submitted by #0331.

*AG4393	Fibroblast, lung	Lung	00 04 00	F		12	>80	EEMEM 20U

The culture was initiated on 6/25/79 from explants of minced lung tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XX; normal diploid female. A matched skin-derived fibroblast culture from same fetus is AG4392. Submitted by #0331.

*AG4432	Fibroblast, lung	Lung	00 04 00	F		10	>80	EEMEM 20U

The culture was initiated on 9/26/79 from explants of minced lung tissue taken from a therapeutically aborted fetus of 15 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XX; normal diploid female. A matched skin-derived fibroblast culture from same fetus is AG4431. Submitted by #0331.

AG4452	Fibroblast, lung	Lung	00 04 00	F	BL	11		EEMEM 20U

The culture was initiated on 9/2/80 from explants of minced lung tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. A matched skin-derived fibroblast culture from same fetus is AG4451. Submitted by #0331.

AG6562	Fibroblast, lung	Lung	00 04 00	F	WH	10		EEMEM 20U

The culture was initiated on 12/2/82 from explants of minced lung tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. A matched skin-derived fibroblast culture from same fetus is AG6561. Culture submitted by #0331.

AG4526	Fibroblast, lung	Lung	00 04 15	M		10		EEMEM 20U

The culture was initiated on 5/11/78 from explants of minced lung tissue taken from a therapeutically aborted fetus of 17 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to the Repository is 46,XY; normal diploid male. A matched skin-derived fibroblast culture from same fetus is AG4525. Culture submitted by #0331.

AG6558	Fibroblast, lung	Lung	00 04 15	M	BL	11		EEMEM 20U

The culture was initiated on 10/05/82 from explants of minced lung tissue taken from a therapeutically aborted fetus of 18 weeks gestational age. The cell morphology is fibroblast-like. A matched skin-derived fibroblast culture from same fetus is AG6557. Culture submitted by #0331.

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	PDL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Fetal Tissue							
AG6560	Fibroblast, lung	Lung	00 04 21	M	BL	11		EEMEM 20U

The culture was initiated on 10/15/82 from explants of minced lung tissue taken from a therapeutically aborted fetus of 19 weeks gestational age. The cell morphology is fibroblast-like. A matched skin-derived fibroblast culture from same fetus is AG6559. Culture submitted by #0331.

*AG4434	Fibroblast, lung	Lung	00 05 00	M		11		EEMEM 20U

The culture was initiated on 1/15/80 from explants of minced lung tissue taken from a therapeutically aborted fetus of 19 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XY; normal diploid male. A matched skin-derived fibroblast culture from same fetus is AG4433. Submitted by #0331.

AG6556	Fibroblast, lung	Lung	00 05 00	M	WH	12		EEMEM 20U

The culture was initiated on 2/11/82 from explants of minced lung tissue taken from a therapeutically aborted fetus of 20 weeks gestational age. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. A matched skin-derived fibroblast culture from same fetus is AG6555. Culture submitted by #0331.

*AG4449	Fibroblast, skin	Buttock-thigh	00 03 00	M		11		EEMEM 20U

The culture was initiated on 5/20/80 from explants of minced skin tissue taken from a therapeutically aborted fetus of 12 weeks gestational age. The cell morphology is fibroblast-like. A matched lung-derived fibroblast culture from same fetus is AG4450. Submitted by #0331.

*AG4392	Fibroblast, skin	Buttock-thigh	00 04 00	F		11	>80	EEMEM 20U

The culture was initiated on 6/25/79 from explants of minced skin tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XX; normal diploid female. A matched lung-derived fibroblast culture from same fetus is AG4393. Submitted by #0331.

*AG4431	Fibroblast, skin	Buttock-thigh	00 04 00	F		11	>80	EEMEM 20U

The culture was initiated on 9/26/79 from explants of minced skin tissue taken from a therapeutically aborted fetus of 15 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XX; normal diploid female. A matched lung-derived fibroblast culture from same fetus is AG4432. Submitted by #0331.

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	PDL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Fetal Tissue							
AG4451	Fibroblast, skin	Buttock-thigh	00 04 00	F	BL	13		HF10 20U

The culture was initiated on 9/2/80 from explants of minced skin tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A matched lung-derived fibroblast culture from same fetus is AG4452. Submitted by #0331.

*AG4433	Fibroblast, skin	Buttock-thigh	00 05 00	M		12		EEMEM 20U

The culture was initiated on 1/15/80 from explants of minced skin tissue taken from a therapeutically aborted fetus of 19 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to Repository is 46,XY; normal diploid male. A matched lung-derived fibroblast culture from same fetus is AG4434. Submitted by #0331.

*AG4525	Fibroblast, skin	Buttock	00 04 15	M		9		EEMEM 20U

The culture was initiated on 5/11/78 from explants of minced skin tissue taken from a therapeutically aborted fetus of 17 weeks gestational age. The cell morphology is fibroblast-like. The karyotype determined prior to submission to the Repository is 46,XY; normal diploid male. A matched lung-derived fibroblast culture from same fetus is AG4526. Culture submitted by #0331.

AG6561	Fibroblast, skin	Lower back	00 04 00	F	WH	12		EEMEM 20U

The culture was initiated on 12/2/82 from explants of minced skin tissue taken from a therapeutically aborted fetus of 16 weeks gestational age. The cell morphology is fibroblast-like. A matched lung-derived fibroblast culture from same fetus is AG6562. Culture submitted by #0331.

AG6557	Fibroblast, skin	Lower back	00 04 15	M	BL	11		EEMEM 20U

The culture was initiated on 10/5/82 from explants of minced skin tissue taken from a therapeutically aborted fetus of 18 weeks gestational age. The cell morphology is fibroblast-like. A matched lung-derived fibroblast culture from same fetus is AG6558. Culture submitted by #0331.

AG6559	Fibroblast, skin	Lower back	00 04 21	M	BL	10		HEMEM 20U

The culture was initiated on 10/15/82 from explants of minced skin tissue taken from a therapeutically aborted fetus of 19 weeks gestational age. The cell morphology is fibroblast-like. A matched lung-derived fibroblast culture from same fetus is AG6560. Culture submitted by #0331.

AG6555	Fibroblast, skin	Lower back	00 05 00	M	WH	12		EEMEM 20U

The culture was initiated on 2/11/82 from explants of minced skin tissue taken from a therapeutically aborted fetus of 20 weeks gestational age. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. A matched lung-derived fibroblast culture from same fetus is AG6556. Culture submitted by #0331.

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REPO	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	POL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Non-Fetal Tissue							
AG2101	Fibroblast, endometrial	Uterus	27 00 00	F				EEMEM 10U
	The culture was initiated using explants of endometrial tissue obtained from a uterine curettage procedure performed 5/17/76. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at passage 8 and submitted by #0203.							
AG2102	Fibroblast, endometrial	Uterus	27 00 00	F		8		HEMEM 20U
	The culture was initiated using explants of endometrial tissue obtained from a uterine curettage procedure performed 1/10/77. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture submitted by #0203.							
AG2603	Fibroblast, lung	Lung	35 00 00	F		3		EEMEM 20U
	The biopsy was taken post-mortem on 8/4/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A matched skin-derived fibroblast culture from same donor is AG2602. Biopsy submitted by #0200.							
*AG2258	Fibroblast, lung	Lung	46 00 00	F	WH	2		EEMEM 20U
	The biopsy was taken 5 hr. post-mortem on 3/17/76. Cause of death was coronary artery disease. The culture was initiated using explants of minced lung tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female, and 18% of cells examined had random chromosomal abnormalities. Biopsy submitted by #0200.							
*AG2262	Fibroblast, lung	Lung	61 00 00	M	WH	4		EEMEM 20U
	The biopsy was taken 20 hr. post-mortem on 3/18/77. Cause of death was coronary artery disease. The culture was initiated using explants of minced lung tissue. The karyotype is 46,XY; normal diploid male. A matched skin-derived fibroblast culture from same donor is AG2261A. Biopsy submitted by #0200.							
*AG1437A	Fibroblast, skin	Foreskin	00 00 03	M	WH	11		EEMEM 20U
	The culture was initiated on 12/22/75 from explants of minced foreskin removed ante-mortem. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.							
*AG1439	Fibroblast, skin	Foreskin	00 00 03	M	BL	5		EEMEM 20U
	The culture was initiated on 12/23/75 from explants of minced foreskin removed ante-mortem 1 day earlier. The cell morphology is fibroblast-like. The karyotype is 46,XY,21s+; normal diploid male with a polymorphism. Culture submitted by #0152.							

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	POL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Non-Fetal Tissue							
AG1440	Fibroblast, skin	Foreskin	00 00 03	M	BL	5		EEMEM 20U

The culture was initiated on 12/23/75 from explants of minced foreskin removed ante-mortem 1 day earlier. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male with 20% of cells showing random chromosomal abnormalities. Culture submitted by #0152.

*AG1518A	Fibroblast, skin	Foreskin	00 00 03	M		10		EEMEM 20U

The culture was initiated on 1/21/76 from explants of minced foreskin removed ante-mortem 1 day earlier. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

*AG1519	Fibroblast, skin	Foreskin	00 00 03	M		3		EEMEM 20U

The culture was initiated on 1/21/76 from explants of minced foreskin removed ante-mortem 1 day earlier. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

AG1520	Fibroblast, skin	Foreskin	00 00 03	M		3		EEMEM 20U

The culture was initiated on 1/21/76 from explants of minced foreskin removed ante-mortem 1 day earlier. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

*AG1521	Fibroblast, skin	Foreskin	00 00 03	M		4		EEMEM 20U

The culture was initiated on 1/21/76 from explants of minced foreskin removed ante-mortem. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

*AG1522A	Fibroblast, skin	Foreskin	00 00 03	M		9		EEMEM 20U

The culture was initiated on 1/21/76 from explants of minced foreskin removed ante-mortem. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

*AG1523	Fibroblast, skin	Foreskin	00 00 03	M		5		EEMEM 20U

The culture was initiated 1/21/76 from explants of minced foreskin removed ante-mortem. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture submitted by #0152.

AG7095	Fibroblast, skin	Foreskin	02 00 00	M	BL	3		EEMEM 20U

The culture was initiated on 10/14/83 from explants of minced foreskin removed ante-mortem 4 hr. earlier. The cell morphology is fibroblast-like. Culture submitted by #0414.

AGING CELL REPOSITORY CULTURES

APPARENTLY NORMAL HUMAN FIBROBLASTS

REP#	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	RACE	PDL FROZEN	EST. IN VITRO LIFE SPAN	MEDIA
	Apparently Normal: Non-Fetal Tissue							
AG6103	Fibroblast, skin	Foreskin	29 07 03	M	BL	5		EEMEM 20U

The culture was initiated 10/11/82 from an ante-mortem, surgical specimen obtained 4 hours earlier. Minced tissue explants were used for the primary. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Biopsy submitted by #0442.

AG6858	Fibroblast, skin	Foreskin	47 00 00	M	BL	3		EEMEM 20U

The culture was initiated 5/13/83 from an ante-mortem, surgical specimen obtained 3 hr. earlier. Minced tissue explants were used for the primary. The cell morphology is fibroblast-like. Biopsy submitted by #0465.

*AG2602	Fibroblast, skin	Abdomen	35 00 00	F		3		EEMEM 20U

The biopsy was taken post-mortem on 8/4/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. A matched lung-derived fibroblast culture from same donor is AG2603. Biopsy submitted by #0200.

*AG2222	Fibroblast, skin	Abdomen	49 00 00	M	WH	3		EEMEM 20U

The biopsy was obtained 2 hr. post-mortem on 3/8/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male, and 34% of cells examined had random chromosomal abnormalities. Biopsy submitted by #0200.

*AG2261A	Fibroblast, skin	Abdomen	61 00 00	M	WH	9		EEMEM 20U

The biopsy was taken 20 hr. post-mortem on 3/18/77. Cause of death was coronary artery disease. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. A matched lung-derived fibroblast culture from same donor is AG2262. Biopsy submitted by #0200.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Alzheimer Disease of Brain - 10430

*AG5770 Fibroblast, skin 70 00 00 M WH Calf +- PROBAND

The donor presented a 7 and 1/2 year history of progressive mental deterioration typical of senile dementia of the Alzheimer type. At autopsy, Alzheimer plaques were observed in the hippocampus and temporal cortex along with neurofibrillary tangles and Hirano bodies. The biopsy was taken post-mortem from skin of the right calf. The culture was initiated on 3/8/82 using explants of minced tissue. The cell morphology is fibroblast-like, and the karyotype is 46,XY; normal diploid male. The culture is frozen at PDL 7 and was submitted by #0421. Culture medium: EEMEM 20U.

AG6262 Fibroblast, skin 65 07 22 M WH Arm +- PROBAND

The donor had a history of progressive dementia with memory deficiencies, requiring hospitalization at age 62 years. A CT scan at age 59 showed nonfocal cortical atrophy. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 1/20/83 using explants of minced tissue. The cell morphology is fibroblast-like. The culture is a mosaic with karyotype: 46,XY,9qh+/46,XY,9qh+,t(4;9)(4pter>4q21::9q34>9qter;9pter>9q34::4q21>4qter), balanced; 44%/48%, and 8% of cells show random chromosomal abnormalities. Culture frozen at PDL 6. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

AG6263 Fibroblast, skin 67 00 00 F WH Arm +- PROBAND

The donor had a history of dementia and behavioral changes since age 60 years. A CT scan revealed mild central and moderate peripheral cortical atrophy. The skin biopsy was taken ante-mortem from the upper left arm. Culture initiated on 1/20/83 using explants of minced skin. The cell morphology is fibroblast-like, & the karyotype is 46,XX; normal diploid female. Culture frozen at PDL 4. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

*AG6264 Fibroblast, skin 62 00 00 F Arm +- PROBAND

Donor has history of presenile dementia. A cranial CT scan showed a mild diffuse cerebral atrophy. Both parents of donor showed signs of senile dementia. The skin biopsy was taken ante-mortem from the upper left arm. Culture was initiated 1/20/83 using explants of minced skin. The cell morphology is fibroblast-like. The karyotype is 46,XX; and 30% of cells examined showed random chromosomal abnormalities. Culture frozen at PDL 5. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

*AG6265 Fibroblast, skin 61 00 00 M WH Arm +- PROBAND

The donor has a history of progressive presenile dementia. His father had a five year progressive dementing illness ending in death at age 66. The skin biopsy was taken ante-mortem from the upper left arm. Culture was initiated on 1/20/83 using explants of minced skin. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 4. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Alzheimer Disease of Brain - 10430

AG7374	Fibroblast, skin	71 02 19	M	WH	Arm	+-	PROBAND		

The donor had a history of progressive dementia including loss of memory, confusion and disorientation, eventually requiring institutional placement. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 2/28/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

AG7375	Fibroblast, skin	71 00 00	M	WH	Arm	+-	PROBAND		

The donor had a 9 year history of progressive mental deterioration. A CT scan at age 67 showed marked diffuse cortex cerebral atrophy. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 2/29/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

AG7376	Fibroblast, skin	59 05 04	M	WH	Arm	+-	PROBAND		

The donor had a 6 year history of progressive intellectual deterioration. A CT scan showed severe cerebral atrophy. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 2/29/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

AG7377	Fibroblast, skin	59 05 21	M	WH	Arm	+-	PROBAND		

The donor had a 9 year history of progressive mental deterioration and development of aggressive behavior. A CT scan revealed moderate to marked cortical atrophy. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 2/29/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. Biopsy submitted by #0451. Culture medium: EEMEM 20U.

*AG0364B	Fibroblast, skin	53 09 28	M	WH	Arm	+-	PROBAND	87	1

The donor showed signs of a progressive dementia as early as age 42. Many members of the donor's family are affected, and the pattern of inheritance of the dementia is autosomal dominant. The skin biopsy was taken ante-mortem on 1/9/74 from the upper arm. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. A fibroblast culture from an unaffected niece is GM0490. Culture frozen at PDL 3 and submitted by #0139. Culture medium: EEMEM 20U.

*AG4401A	Fibroblast, skin	53 00 00	F	WH	Skin	+-	PROBAND	747	1

The donor is a clinically affected member of a family with Alzheimer disease. She has a 5 year history of severe dementia, decreased short-term memory and decreased word finding ability. A brain biopsy confirmed the diagnosis. Fibroblast cultures from 2 affected sibs are AG4400 & AG4402. The skin biopsy for culture AG4401A was taken ante-mortem. The culture was initiated on 10/17/80 using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 9. Biopsy submitted by #0305. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Alzheimer Disease of Brain - 10430

*AG4400	Fibroblast, skin	61 00 00	F	WH	Skin	+-	AFF SIS	747	2

The donor is a clinically affected member of a family with Alzheimer disease. She has a 5 year history of abnormal mental status, including disorientation, aphasia and absence of memory, but no spasticity. Fibroblast cultures from 2 affected sibs are AG4401A and AG4402. The biopsy was taken ante-mortem. The culture AG4400 was initiated on 10/17/80 using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is a mosaic with a karyotype 46,XX/46,XX,t(1;7)(1pter>1q25::7p15>7pter;7qter>7p15::1q25>1qter), balanced; 88%/12%. Culture frozen at PDL 7. Biopsy submitted by #0305. Culture medium: EEMEM 20U.

AG4402	Fibroblast, skin	47 00 00	M	WH	Arm	+-	AFF BRO	747	3

The donor is a clinically affected member of a family with Alzheimer disease, with signs of early dementia, inappropriate jocularity and occasional paraphasias. Fibroblast cultures from 2 affected sibs are AG4400 & AG4401A. The biopsy was taken ante-mortem from skin of the right forearm. The culture was initiated on 10/17/80 using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 5. Biopsy submitted by #0305. Culture medium: EEMEM 20U.

*AG5810A	Fibroblast, skin	79 00 00	F	WH	Arm	+-	PROBAND	907	1

The donor is clinically affected with severe late stage dementia, typical of Alzheimer disease. Three sibs died with autopsy confirmed Alzheimer disease. The biopsy was taken ante-mortem from skin of the forearm, and the culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture submitted by #0421. Culture medium: EEMEM 20U.

AG5809	Fibroblast, skin	63 00 00	F	WH	Skin	+-	AFF SIS	907	2

The donor is clinically affected with moderate dementia of insidious onset. Three sibs died with autopsy proven Alzheimer disease. The culture was initiated with explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is a mosaic with karyotype 46,XX/47,XX,+21/47,XX,t(4;17)(4pter>4q29::17q25>17qter;17pter>17q25::4q29>4qter); 52%/44%/4%. The culture is frozen at PDL 6 and was submitted by #0421. Culture medium: EEMEM 20U.

AG6204	Lymphoblast	67 00 00	M	WH	Blood	+-	PROBAND	922	1

The donor showed severe dementia, progressive memory loss, word finding difficulty, dyscalculia and mild to moderate cerebral atrophy on CAT scan. There is no family history of Alzheimer disease. The culture was initiated on 11/24/82 by transformation of lymphocytes with EB virus. The cells grow in suspension, and their morphology is spherical. Blood specimen submitted by #0441. Culture medium: R1640 15I.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Alzheimer Disease of Brain - 10430

AG6205 Fibroblast, skin — 67 00 00 — M — WH — Skin — +- — PROBAND — 922 — 1

The donor showed severe dementia, progressive memory loss, word finding difficulty, dyscalculia and mild to moderate cerebral atrophy on CAT scan. There is no family history of Alzheimer disease. The culture was initiated on 11/23/82 using explants of minced skin tissue taken ante-mortem. The morphology is fibroblast-like. The culture is a mosaic with the karyotype 46,XY/46,XY,+p+,12q-/46,XY,t(2;8)(p23;q13)balanced: 70%/14%/10%/6% with random chromosomal abnormalities. Biopsy submitted by #0441. Culture medium: EEMEM 20U.

AG6868 Lymphoblast — 60 01 25 — F — WH — Blood — +- — PROBAND — 963 — 1

The donor has a history of moderate dementia, impaired memory, visuo-spacial disorientation, dyscalculia, a diffuse EEG and cortical atrophy on CAT scan. There is no family history of Alzheimer disease. The culture was initiated on 5/19/83 by transformation of lymphocytes with EB virus. The cells grow in suspension, and their morphology is spherical. Blood specimen submitted by #0363. Culture medium: R1640 15I.

AG6869 Fibroblast, skin — 60 01 25 — F — WH — Skin — +- — PROBAND — 963 — 1

The donor has a history of moderate dementia, impaired memory, visuo-spacial disorientation, dyscalculia, a diffuse EEG and cortical atrophy on CAT scan. There is no family history of Alzheimer disease. The culture was initiated on 5/19/83 using explants of minced skin tissue taken ante-mortem. The cell morphology is fibroblast-like, and the karyotype is 46,XX; normal diploid female. Culture frozen at PDL 5. Biopsy submitted by #0363. Culture medium: EEMEM 20U.

Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890

AG2496 Fibroblast, skin — 13 00 00 — M — — Skin — -- — PROBAND

Donor was diagnosed in U.S.S.R. The cell morphology is fibroblast-like. The karyotype is 46,XY with random chromosome abnormalities in 65% of examined cells. Culture submitted by #0196. Culture medium: EEMEM 20U.

AG4405 Fibroblast, skin — 07 05 10 — M — WH — Arm — -- — PROBAND

Donor is clinically affected, with a negative family history for ataxia telangiectasia. The biopsy was taken ante-mortem from skin of the left forearm on 10/23/80. The culture was initiated using explants of minced skin tissue. Cell morphology is fibroblast-like. The karyotype is 46,XY with random chromosomal abnormalities in 50% of examined cells. Culture frozen at PDL 4. Biopsy submitted by #0407. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Ataxia-Telangiectasia (Louis-Bar Syndrome) - 20890

*AG5749A	Lymphoblast	10 00 00	M	WH	Blood	--	PROBAND		

Donor displayed symptoms of telangiectasia in eyes, vitiligo, unsteady gait, absence of IgA and possible cerebellar degeneration. The culture was established spontaneously from a peripheral blood specimen. Karyotype before submission to Repository was 46,XY; normal diploid male. Culture submitted by #0346. Culture medium: R1640 15I.

*AG3058	Fibroblast, skin	14 02 14	F	BL	Skin	--	PROBAND	493	1

The donor is clinically affected with ataxia-telangiectasia and also with acute leukemia. Clinical manifestations are gross truncal and limb ataxia, cafe au lait spots, ocular telangiectasia and generalized lymphadenopathy. One sister died with A-T. The skin biopsy was taken ante-mortem on 6/9/78. The culture was initiated using explants of minced tissue. It shows an increased sensitivity to cell killing by X-irradiation. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0296. Culture medium: EEMEM 20U.

*AG3057	Fibroblast, skin	46 08 24	F	BL	Skin	+-(0)	MOTHER	493	2

Donor is a clinically unaffected mother with two affected daughters. The skin biopsy was taken ante-mortem on 6/9/78. The culture was initiated using explants of minced tissue. It shows a normal sensitivity to cell killing by X-irradiation. The cell morphology is fibroblast-like. Culture frozen at PDL 6. Biopsy submitted by #0296. Culture medium: EEMEM 20U.

*AG3059	Fibroblast, skin	47 08 00	M	BL	Skin	+-(0)	FATHER	493	3

Donor is a clinically unaffected father with two affected daughters. The skin biopsy was taken ante-mortem on 6/12/78. The culture was initiated using explants of minced tissue. It shows a normal sensitivity to cell killing by X-irradiation. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0296. Culture medium: EEMEM 20U.

Bloom Syndrome - 21090

AG6040	Fibroblast, skin	27 00 00	F	WH	Skin	--	PROBAND		

Donor is clinically affected and of Ashkenazi background. Clinical symptoms include short stature, elongated facies and adenocarcinoma of colon. Biopsy was taken ante-mortem on 11/21/80. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. An increased frequency of sister chromatid exchange was found in cells and confirms diagnosis. The culture is frozen at PDL 11 and was submitted by #0077. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Cockayne Syndrome - 21640

AG5012	Fibroblast, skin	11 00 00	F	WH	Skin	--	PROBAND		

The donor has clinical symptoms of short stature, mental deficiency, ataxia, retinal degeneration, senile-like changes, and a positive family history. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture submitted by #0395. Culture medium: EEMEM 20U.

AG5243A	Fibroblast, skin	01 02 18	F	WH	Skin	--	PROBAND		

The donor displays clinical symptoms of severe growth retardation, mental retardation, microphthalmia, spasticity of upper and lower extremities, and peripheral neuropathy. The biopsy was taken ante-mortem on 10/30/81 from skin on the left peroneal region. The culture was initiated using explants of minced tissue. The karyotype is 46,XX; normal diploid female. Biopsy submitted by #0305. Culture medium: EEMEM 20U.

*AG6244	Fibroblast, skin	04 00 00	F	WH	Skin	--	PROBAND		

Donor displayed clinical symptoms of growth failure, short stature, typical facies, photosensitivity, slow nerve conduction velocities, putamenal calcifications and senile-like changes. The culture was initiated on 10/20/82. The morphology is fibroblast-like. Culture submitted by #0488. Culture medium: EEMEM 20U.

AG6269	Fibroblast, skin	07 09 19	M	WH	Skin	--	PROBAND		

The donor displayed clinical symptoms of postnatal short-stature, photosensitivity, corneal dystrophy, hyper- and hypopigmentations of the skin and learning difficulties. The biopsy was taken ante-mortem on 9/29/82. The culture was initiated from explants of minced skin tissue. The morphology is fibroblast-like. Culture submitted by #0078. Culture medium: HEMEM 20U.

AG7946	Fibroblast, skin	06 09 01	M	BL	Arm	--	PROBAND		

The donor has symptoms of dwarfism, mental retardation, orbital fat atrophy, optic atrophy, pigmentary retinal changes and facies characteristic for Cockayne syndrome. The culture was initiated on 11/2/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at PDL 11 and was submitted by #0309. Culture medium: EEMEM 20U.

AG7075	Lymphoblast	10 10 07	F	WH	Blood	--	PROBAND	989	1

The donor has clinical symptoms of photophobia, dwarfism, mental retardation, cataracts, retinopathy and optical atrophy. The culture was initiated on 9/23/83 by transformation of lymphocytes with EB virus. The cells grow in suspension, and their morphology is spherical. Blood specimen submitted by #0482. Culture medium: R1640 15I.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#
		HUMAN CELL CULTURES: AGING							
	Cockayne Syndrome - 21640								
AG7076	Fibroblast, skin	10 10 07	F	WH	Skin	--	PROBAND	989	1
	Donor has clinical symptoms of photophobia, dwarfism, mental retardation, cataracts, retinopathy and optical atrophy. The skin biopsy was taken ante-mortem on 9/22/83. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0482. Culture medium: EEMEM 20U.								
	Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585								
AG6064	Fibroblast, skin	34 00 00	M	WH	Skin		PROBAND	906	1
	Donor was diagnosed with Type II diabetes, MODY, at age 25 years. His GTTs fluctuate between impaired glucose tolerance, diabetes and normal. His father, paternal aunt and 3 sibs are also affected. The skin biopsy was taken ante-mortem on 9/10/82. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 6. Biopsy submitted by #0034. Culture medium: EEMEM 20U.								
AG6062	Fibroblast, skin	26 00 00	F	WH	Skin		SIS	906	2
	Donor is at risk for developing Type II diabetes, MODY. GTTs were normal and nondiagnostic (close to impaired). She has a positive family history, and her father, paternal aunt and 3 sibs are affected. The skin was taken ante-mortem on 9/10/82. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0034. Culture medium: EEMEM 20U.								
AG6063	Fibroblast, skin	61 00 00	F	WH	Skin		AFF AUNT	906	3
	Donor was diagnosed with Type II insulin-requiring diabetes at age 48 years. She has a positive family history with many affected members. The skin biopsy was taken ante-mortem on 9/10/82. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 3. Biopsy submitted by #0034. Culture medium: EEMEM 20U.								
AG6083	Fibroblast, skin	35 00 00	M	WH	Skin		PROBAND	913	1
	Donor was diagnosed with Type II diabetes, MODY, at age 23 years. His GTTs are consistently diabetic. He has a positive family history with both parents and 4 sibs also affected. The culture was initiated on 9/30/82 using explants of minced tissue taken ante-mortem. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0034. Culture medium: EEMEM 20U.								

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Diabetes Mellitus: Maturity Onset Diabetes - 22210 & 12585

AG6084A	Fibroblast, skin	41 00 00	M	WH	Skin		AFF BRO	913	2

Donor was diagnosed with Type II diabetes at age 29 years. He has a positive family history with both parents and 4 sibs also affected. The culture was initiated on 9/30/82 using explants of minced skin tissue taken ante-mortem. The cell morphology is fibroblast-like. Biopsy submitted by #0034. Culture medium: EEMEM 20U.

AG6085	Fibroblast, skin	44 00 00	M	WH	Skin		AFF BRO	913	3

Donor was diagnosed with Type II diabetes, MODY, at age 35 years. He has a positive family history with both parents and 4 sibs also affected. The culture was initiated on 9/30/82 using explants of minced skin tissue taken ante-mortem. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0034. Culture medium: EEMEM 20U.

AG6086	Fibroblast, skin	50 00 00	F	WH	Skin		AFF SIS	913	4

Donor was diagnosed with Type II diabetes at age 41 years. She has a positive family history with both parents & 4 sibs also affected. The culture was initiated on 9/30/82 using explants of minced skin tissue taken ante-mortem. The cell morphology is fibroblast-like. Culture frozen at PDL 6. Biopsy submitted by #0034. Culture medium: EEMEM 20U.

AG6961	Fibroblast, skin	50 00 00	M	WH	Arm		AFF BRO	913	5

Donor is at risk for developing Type II diabetes and has been diagnosed with impaired glucose tolerance. He has a positive family history for MODY with both parents and 4 sibs affected. The biopsy was taken ante-mortem from skin of right forearm. The culture was initiated on 8/23/83 using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0034. Culture medium: EEMEM 20U.

Dyskeratosis Congenital Zinsser-Cole-Engman Syndrome - 30500

AG3739A	Lymphoblast	20 07 26	M	WH	Blood	Y-	PROBAND	547	1

Donor is clinically affected and presented symptoms of reticulated cutaneous hyperpigmentation, telangiectasia, atrophy, nail dystrophy and squamous cell carcinoma of tongue. The culture was initiated on 1/4/80 by transformation of lymphocytes with EB virus. The cells grow in suspension and their morphology is spherical. Blood specimen submitted by #0295. Culture medium: R1640 20I.

AG3738A	Lymphoblast	42 05 22	F	WH	Blood	+-(D)	MOTHER	547	2

Donor is a clinically unaffected obligate carrier with an affected son. The culture was initiated on 3/11/80 by transformation of lymphocytes with EB virus. The cells grow in suspension and their morphology is spherical. Blood specimen submitted by #0295. Culture medium: R1640 15I.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Dyskeratosis Congenita; Zinsser-Cole-Engman Syndrome - 30500

AG4645	Fibroblast, skin	11 00 00	M	WH	Involved skin	Y-	PROBAND	758	1

Donor is affected and presented symptoms of skin eruptions, anemia, nail dystrophy and esophageal abnormalities. Family history is negative. The biopsy was taken ante-mortem from a skin eruption (involved area). The culture was initiated on 2/21/81 using explants of minced tissue. The morphology is fibroblast-like. Culture frozen at PDL 3. Biopsy submitted by #0199. Culture medium: EEMEM 20U.

*AG4646	Fibroblast, skin	11 00 00	M	WH	Uninvolved skin	Y-	PROBAND	758	1

Donor is affected and presented symptoms of skin eruptions, anemia, nail dystrophy and esophageal abnormalities. Family history is negative. The biopsy was taken ante-mortem from uninvolved skin. The culture was initiated on 2/21/81 using explants of minced tissue. The cell morphology is fibroblast-like. The culture is sensitive to cell killing by mitomycin-C. Culture frozen at PDL 4. Biopsy submitted by #0199. Culture medium: EEMEM 20U.

Lipoatrophic Diabetes (Seip Syndrome, Berardinelli Syndrome) - 26970

*AG0990B	Fibroblast, skin	15 00 00	M	WH	Skin	--	PROBAND	797	1

Donor displayed symptoms of hypertriglyceridemia and an abnormal glucose tolerance test. He was originally diagnosed with atypical progeria. The culture was initiated on 8/27/73 from explants of skin taken ante-mortem and was designated KH2. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male and 25% of examined cells showed chromatid or chromosome breakage. The culture is frozen at approximate PDL 15 and was submitted by #0055. Culture medium: EEMEM 20U.

Mulibrey Nanism - 25325

AG2122	Fibroblast, skin	10 05 05	F		Skin	--	PROBAND		

The biopsy was taken ante-mortem on 1/31/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture is frozen at PDL 13. Biopsy submitted by #0004. Culture medium: EEMEM 20U.

AG2505	Fibroblast, skin	05 10 11	F	WH	Skin	--	PROBAND		

The biopsy was taken 6/20/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture is frozen at PDL 5. Biopsy submitted by #0004. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Mulibrey Nanism - 25325

AG2506 Fibroblast, skin 09 04 15 F WH Skin -- PROBAND

The biopsy was taken 6/20/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture is frozen at PDL 4. Biopsy submitted by #0004. Culture medium: EEMEM 20U.

Myotonic Dystrophy (Steinert Disease) - 16090

AG4475 Fibroblast, skin 57 00 00 M WH Arm +- PROBAND

The donor had symptoms of myotonia, weakness, testicular atrophy and gynecomastia. He did not have cataracts or frontal balding. The biopsy was taken ante-mortem on 11/18/80 from the skin of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture is frozen at PDL 3. Biopsy submitted by #0210. Culture medium: EEMEM 20U.

Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667

AG7091 Lymphoblast 28 00 09 F WH Blood +- PROBAND

The donor displays the following clinical signs of progeria: short stature, alopecia, severe osteoporosis with resorption of clavicles and upper ribs, multiple fractures, typical facies, and dystrophic nails with resorption of distal phalanges. Unusual features are absence of coronary artery occlusions at age 28, and monthly menses since age 15. Parents and seven sibs are clinically unaffected. The blood specimen was obtained ante-mortem. The culture was initiated on 10/12/83 by transformation with EB virus. The culture grows in suspension, and the cell morphology is spherical. The karyotype is 46,XX; normal diploid female. Blood specimen submitted by #0470. Culture medium: R1640 15I.

AG0989B Fibroblast, skin 20 00 00 M Skin +- PROBAND

Donor was diagnosed with atypical progeria and an unspecified type of cachectic dwarfism. The culture was initiated on 2/11/69 using explants of minced skin and was designated Strain S.J. The cell morphology is fibroblast-like. Culture is frozen at PDL 22 and was submitted by #0055. Culture medium: EEMEM 20U.

AG1178 Fibroblast, skin 20 00 00 M Skin +- PROBAND

Donor had clinical signs of small eyes, spontaneous nystagmus, increased corneal curvature, absence of teeth, severe micrognathia, small testes (3ml), diminished muscular mass, absence of body hair, and an old looking face with a depressed nose. Scalp hair is sparse and thin, not greyish. Donor died at age 34 years. Culture was obtained from the Canadian Cell Repository as Strain WG380, submitted by #0049. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#
		HUMAN CELL CULTURES: AGING							
	Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667								
*AG1972A	Fibroblast, skin	13 10 03	F	WH	Thoracic skin	+-	PROBAND		
	Donor showed classic features of progeria. The biopsy was taken ante-mortem on 2/14/75 from skin on the thorax area. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 46, XX; normal diploid female with 6% of examined cells showing random chromosome loss. Culture frozen at PDL 22. Culture submitted by #0091. Culture medium: EEMEM 20U.								
*AG6297	Fibroblast, skin	08 02 04	M	WH	Thigh	+-	PROBAND		
	The donor had classical signs of progeria, and died at age 14 years of congestive heart failure. Autopsy revealed severe coronary-vascular disease. The biopsy was taken ante-mortem on 1/11/72 from skin on the anterior thigh region. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like, and the karyotype is 46,XY; normal diploid male. Culture is frozen at PDL 26, & has a maximum life-span of 40 PD. Culture submitted by #0453. Culture medium: EEMEM 20U.								
*AG6831	Fibroblast, skin	16 08 18	F	WH	Abdomen	+-	PROBAND		
	Donor has classic symptoms of progeria including short stature, alopecia and atherosclerosis. The biopsy was taken post-mortem on 4/25/83 from skin of the thoraco-abdominal area. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture is frozen at PDL 7. Biopsy submitted by #0309. Culture medium: EEMEM 20U.								
AG7493	Fibroblast, skin	01 09 00	F	WH	Arm	+-	PROBAND		
	The donor had physical features of Hutchinson-Gilford syndrome including low weight and alopecia. The skin biopsy was taken ante-mortem from the upper left arm. The culture was initiated on 4/17/84 using explants of minced skin tissue. The cell morphology is fibroblast-like, and the karyotype is 46,XX; normal diploid female. Culture frozen at PDL 5. Biopsy submitted by #0309. Culture medium: EEMEM 20U.								
AG3506	Lymphoblast	13 00 00	M	WH	Blood	+-	PROBAND	172	1
	Donor has classical symptoms of progeria, but no signs of heart disease. His parents and sibs are clinically unaffected. The culture was initiated on 5/16/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. The karyotype is 46,XY; normal diploid male. The blood specimen was obtained ante-mortem and was submitted by #0309. Culture medium: R1640 20I.								
*AG3513	Fibroblast, skin	13 00 00	M	WH	Arm	+-	PROBAND	172	1
	Donor has classical symptoms of progeria, but no signs of heart disease. His parents and sibs are clinically unaffected. The biopsy was taken ante-mortem on 5/15/79 from skin on the medial forearm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. An increased thermolabile component of triosephosphate isomerase has been reported for this culture. Culture frozen at PDL 6. Biopsy submitted by #0309. Culture medium: EEMEM 20U.								

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667

AG3504	Lymphoblast	41 00 00	F	WH	Blood	++	MOTHER	172	2

Donor is clinically unaffected. The culture was initiated on 5/16/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. The blood specimen was obtained ante-mortem and was submitted by #0309. Culture medium: R1640 20I.

AG3512	Fibroblast, skin	41 00 00	F	WH	Skin	++	MOTHER	172	2

Donor is clinically unaffected. The culture was initiated on 5/19/79 using explants of minced skin tissue obtained ante-mortem. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 6. Biopsy submitted by #0309. Culture medium: EEMEM 20U.

AG3505	Lymphoblast	37 00 00	M	WH	Blood	++	FATHER	172	3

Donor is clinically unaffected. The culture was initiated on 5/16/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. The blood specimen was obtained ante-mortem and submitted by #0309. Culture medium: R1640 20I.

AG3511	Fibroblast, skin	37 00 00	M	WH	Skin	++	FATHER	172	3

Donor is clinically unaffected. The culture was initiated on 5/18/79 using explants of minced skin tissue taken ante-mortem. The cell morphology is fibroblast-like, and the karyotype is 46,XY; normal diploid male. Culture frozen at PDL 6 and is a poor grower. Biopsy submitted by #0309. Culture medium: EEMEM 20U.

AG3507	Lymphoblast	15 00 00	F	WH	Blood	++	SIS	172	4

Donor is clinically unaffected. The culture was initiated on 5/16/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3508	Lymphoblast	17 00 00	M	WH	Blood	++	BRO	172	5

Donor is clinically unaffected. The culture was initiated on 5/16/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667

AG3345	Lymphoblast	35 00 00	F	WH	Blood	++	MOTHER	543	1

Donor is clinically unaffected. The culture was initiated on 2/9/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3346	Lymphoblast	35 00 00	M	WH	Blood	++	FATHER	543	2

Donor is clinically unaffected. The culture was initiated on 2/8/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

*AG3344	Lymphoblast	10 00 00	M	WH	Blood	+-	PROBAND	543	3

Donor had classical phenotype for progeria and scleroderma as an infant. Family history is negative. The culture was initiated on 2/8/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3343	Lymphoblast	13 00 00	F	WH	Blood	++	SIS	543	4

Donor is clinically unaffected. The culture was initiated on 2/8/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3342	Lymphoblast	11 00 00	M	WH	Blood	++	BRO	543	5

Donor is clinically unaffected. The culture was initiated on 2/8/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

*AG3198	Fibroblast, skin	10 08 11	F	WH	Arm	+-	PROBAND	580	1

Donor had classic symptoms of progeria and also mild maturity onset diabetes. Family history is negative. The biopsy was taken ante-mortem on 10/12/78 from skin on the right dorsal forearm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture is frozen at PDL 6. Biopsy submitted by #0309. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667

*AG3198A	Fibroblast, skin	10 08 11	F	WH	Skin	+-	PROBAND	580	1

Donor had classical symptoms of progeria and also mild maturity onset diabetes. Family history is negative. The biopsy was taken ante-mortem on 10/12/78. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at an approximate PDL of 8 to 12 and was submitted by #0309. Culture medium: EEMEM 20U.

*AG3199	Fibroblast, skin	10 08 11	F	WH	Skin	+-	PROBAND	580	1

Donor had classical symptoms of progeria and also mild maturity onset diabetes. Family history is negative. The biopsy was taken ante-mortem on 10/12/78 from skin on the left lateral hip area. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. The culture is frozen at PDL 4. Biopsy submitted by #0309. Culture medium: EEMEM 20U.

*AG3259A	Lymphoblast	10 10 15	F	WH	Blood	+-	PROBAND	580	1

Donor had classical symptoms of progeria and also had mild maturity onset diabetes. Family history is negative. The culture was initiated on 12/19/78 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. The karyotype is 46,XX; normal diploid female. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3257	Fibroblast, skin	35 00 00	F	WH	Arm	++	MOTHER	580	2

Donor is clinically unaffected. Biopsy was taken ante-mortem from skin on the left dorsal forearm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at approximate PDL 8 to 12 and was submitted by #0309. Culture medium: EEMEM 20U.

AG3260A	Lymphoblast	35 00 00	F	WH	Blood	++	MOTHER	580	2

Donor is clinically unaffected. The culture was initiated on 12/19/78 by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

AG3258	Fibroblast, skin	36 00 00	M	WH	Skin	++	FATHER	580	3

Donor is clinically unaffected. The culture is frozen at approximate PDL 10 to 12. Biopsy submitted by #0309. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#
	HUMAN CELL CULTURES: AGING								
	Progeria (Hutchinson - Gilford Syndrome) - Premature Aging - 17667								
AG3262A	Lymphoblast	06 00 00	F	WH	Blood	++	SIS	580	4
	Donor is clinically unaffected. The culture was initiated on 12/19/78 by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.								
AG3263A	Lymphoblast	12 00 00	M	WH	Blood	++	BRO	580	5
	Donor is clinically unaffected. The culture was initiated on 12/19/78 by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.								
*AG6917	Fibroblast, skin	03 00 09	M	WH	Arm	+-	PROBAND	949	1
	Donor displayed classical signs of progeria including growth retardation; thin skin; a thin, beak-like nose; deficiency of subcutaneous tissue; small clavicles and thin, underdeveloped nails. His parents and 12 sibs are unaffected. Fibroblast cultures from parents are available: AG6298 and AG6299. The biopsy for AG6917 was taken ante-mortem from skin of the anterior forearm. The culture was initiated on 9/6/73 using explants of minced skin. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. The culture is frozen at PDL 28 and was submitted by #0453. Culture medium: EEMEM 20U.								
AG6298	Fibroblast, skin	44 10 22	M	WH	Arm	++	FATHER	949	2
	The donor is clinically normal and father of progeria patient AG6917. The biopsy was taken ante-mortem from the anterior forearm. The culture was initiated on 9/6/73 using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at PDL 20 and was submitted by #0451. Culture medium: EEMEM 20U.								
AG6299	Fibroblast, skin	34 11 21	F	WH	Arm	++	MOTHER	949	3
	The donor is clinically normal and mother of progeria patient AG6917. The biopsy was taken ante-mortem from the anterior forearm. The culture was initiated on 9/6/73 using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at PDL 19 and was submitted by #0451. Culture medium: EEMEM 20U.								
	Rothmund-Thomson Syndrome (Poikiloderma Atrophicans & Cataract) - 26840								
AG3587	Fibroblast, skin	07 04 06	M	WH	Skin	--	PROBAND		
	The donor had symptoms of short stature, pancreatic insufficiency, poikiloderma and telangiectatic erythema of skin, but normal intelligence. The skin biopsy was taken ante-mortem on 7/31/79. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male with random abnormalities in 73% of examined cells. The SCE frequency is normal or near normal. Culture frozen at PDL 4. Biopsy submitted by #0295. Culture medium: EEMEM 20U.								

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Rothmund-Thomson Syndrome (Poikiloderma Atrophicans & Cataract) - 26840

AG5013	Fibroblast, skin	10 00 00	M	WH	Arm	--	PROBAND		

The donor had symptoms of short stature; sparse hair, characteristic facies, poikiloderma, absent right thumb and hyperplastic left thumb. The family history is negative. The biopsy was taken ante-mortem in 1973 from skin on the mesial aspect of the arm. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male with 14% of examined cells showing chromosomal breakage. The culture is frozen at approximate PDL 21 and was submitted by #0395. Culture medium: EEMEM 20U.

AG5139	Fibroblast, skin	10 03 07	M	WH	Lower back	--	PROBAND	818	1

The donor had symptoms of short stature; mental retardation; reticular, pigmented and depigmented atrophic skin lesions; alopecia; cataracts and dystrophic nails. The family history is negative. The biopsy was taken ante-mortem on 8/12/81 from skin of the right sacral area. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0204. Culture medium: EEMEM 20U.

AG5140A	Lymphoblast	10 03 07	M	WH	Blood	--	PROBAND	818	1

The donor had symptoms of short stature; mental retardation; reticular, pigmented and depigmented atrophic skin lesions; alopecia; cataracts and dystrophic nails. The family history is negative. The culture was initiated on 8/17/81 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0204. Culture medium: R1640 15I.

Werner Syndrome - 27770

*AG0780C	Fibroblast, skin	59 11 14	M	WH	Skin	--	PROBAND		

The donor's parents are second cousins. The biopsy was taken ante-mortem on 4/4/80. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is a mosaic with karyotype 46,XY/46,XY,inv(5)(q11q22)/46,XY,inv(5)(q11q22),8q-,9p+,inv(11)(q21q23),15q-,16q+/46,XY with random abnormalities; 16%/38%/30%/16%. Culture frozen at PDL 4. Biopsy submitted by #0126. Culture medium: EEMEM 20U.

AG3141	Fibroblast, skin	29 10 26	F	WH	Skin	--	PROBAND		

The donor had symptoms of premature aging, pigmented and atrophic skin, cataracts and hyperlipidemia type V. The biopsy was taken ante-mortem on 9/20/78. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX with 80% of examined cells having random chromosomal abnormalities. Culture frozen at PDL 2. Biopsy submitted by #0319. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Werner Syndrome - 27770

*AG3364A	Lymphoblast	42 00 00	M	WH	Blood	--	PROBAND		

Donor had symptoms of diabetes, juvenile cataracts, short stature, gray hair, loss of subcutaneous fat, atrophy and ulceration of skin, high-pitched voice and evidence of progeria. The family history is negative for Werner syndrome, diabetes and cataracts. The culture was initiated on 6/1/79 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0309. Culture medium: R1640 20I.

*AG3829	Lymphoblast	42 00 00	M	AI	Blood	--	PROBAND		

The donor had symptoms of diabetes, hypogonadotrophic hypogonadism, cataracts, hyperpigmentation of skin around ankle; gray and thin hair and skin ulcers. The family history is negative. The culture grows in suspension, and the cell morphology is spherical. The karyotype is 46,XY; normal diploid male. The culture was established in 1979 by Dr. E. Henderson and submitted by #0309. Culture medium: R1640 20I.

AG4103	Lymphoblast	37 00 00	F	WH	Blood	--	PROBAND		

The donor had symptoms of diabetes, cataracts, sclerodermatous-like skin, prematurely white hair; non-healing skin ulcers and arteriosclerotic cardiovascular disease. The culture was initiated on 5/23/80 by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. The culture is a mosaic with karyotype 46,XX/46,XX,+9; 36%/64%. Blood lymphocytes submitted by #0309. Culture medium: R1640 15I.

*AG5229	Fibroblast, skin	25 00 00	M	OR	Thigh	--	PROBAND		

The donor had symptoms of short stature, gray hair, hyperpigmentation of skin, juvenile bilateral cataracts, atrophic skin and subcutaneous tissue, and hypogonadism. Diabetes was not detected at time of biopsy. The family history is negative, and the parents are non-consanguineous. The culture was initiated on 10/14/81 from explants of minced skin tissue which had been frozen after biopsy. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,t(12;12)(q13;q24)/46,XY,-4,-21,t(5;8)(q15;q22),+2 mar; 37%/13%/13%/37% with random chromosomal abnormalities. Culture frozen at PDL 3. Biopsy submitted by #0382. Culture medium: EEMEM 20U.

*AG5230	Fibroblast, skin	50 00 00	F	OR	Thigh	--	PROBAND		

The donor had symptoms of short stature, gray hair, bird-like appearance, juvenile bilateral cataracts, atrophic skin and subcutaneous tissue, skin ulcers and diabetes, but not hypogonadism. Family history is negative, but parents are consanguineous. The biopsy was taken ante-mortem and stored frozen. The culture was initiated on 10/14/81 using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is a mosaic with karyotype 46,XX,t(11;11)(p11;q25)/46,XX,t(2;5;13)(q37.1;q11.2p15.3;q14.1),t(6;6)(q21;q27), balanced; 62%/15%/23% with random chromosomal abnormalities. Culture frozen at PDL 3. Biopsy submitted by #0382. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#
		HUMAN CELL CULTURES: AGING							
	Werner Syndrome - 27770								
*AG5232	Fibroblast, skin	48 00 00	M	OR	Thigh	--	PROBAND		

The donor had symptoms of short stature, bird-like appearance, gray hair, juvenile bilateral cataracts, diffuse arteriosclerosis, atrophic skin and subcutaneous tissue, skin ulcers, diabetes and hypogonadism. Family history is positive with an affected mother and sister. Parents are consanguineous. The biopsy was taken ante-mortem and stored frozen. The culture was initiated on 10/14/81 using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male, with 22% of examined cells showing random chromosomal abnormalities including chromosome loss. Culture frozen at PDL 2. Biopsy submitted by #0382. Culture medium: EEMEM 20U.

*AG5233A	Fibroblast, skin	36 00 00	M	OR	Thigh	--	PROBAND		

The donor had symptoms of short stature, bird-like appearance, gray hair, skin hyperpigmentation, juvenile bilateral cataracts, atrophic skin and subcutaneous tissue, diabetes and hypogonadism. The family history is negative, but parents are consanguineous. The biopsy was taken ante-mortem and stored frozen. The culture was initiated on 10/14/81 using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male, with 40% of examined cells showing random chromosomal abnormalities. Culture frozen at PDL 7. Biopsy submitted by #0382. Culture medium: EEMEM 20U.

*AG6300	Fibroblast, skin	36 11 08	M	WH	Arm	--	PROBAND		

The donor developed rapid onset of aging beginning at age 35 years including the development of fine, gray hair, muscle wasting, wrinkling of skin, dystrophic nails, high-pitched voice, hypogonadism and a general aged appearance. The family history is negative, although a sister has developed thin arms and legs. The biopsy was taken ante-mortem 10/25/76 from skin of the anterior forearm. The culture was initiated using explants of skin tissue. The cell morphology is fibroblast-like. The culture is a mosaic with karyotype 46,XY/46,XY,t(7;11)(7qter>7p22::11q23>11qter;11pter>11q23::7p22>7pter), balanced; 88%/12%. Culture frozen at PDL 20. Maximum life-span is 35 to 40 PD. Culture submitted by #0453. Culture medium: EEMEM 20U.

AG6311	Fibroblast, skin	58 00 00	M	OR	Thigh	--	PROBAND		

The donor had symptoms of short stature, gray hair, bird-like appearance, skin hyperpigmentation, juvenile bilateral cataracts, atrophic skin, nails and subcutaneous tissue, skin ulcers, diabetes and hypogonadism. The family history is negative. The biopsy was taken ante-mortem and stored frozen. The culture was initiated on 2/15/83 using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0382. Culture medium: EEMEM 20U.

*AG3593A	Fibroblast, skin	28 00 00	M	WH	Skin	--	PROBAND	546	1

The donor had symptoms of growth retardation, loss of subcutaneous fat, dystrophic nails, hyperkeratosis of palms and soles, cutis marmorata with ulcerations, lank face, baldness, high-pitched voice, bilateral cataracts, and a slightly abnormal GTT. There is no mental retardation or hypogonadism. The family history is positive with a similarly affected brother (AG3594). Unaffected parents are first cousins. The cell morphology is fibroblast-like. The karyotype is 46,XY, t(5;8)(q11;q22)dup(6)(p21>p25),9qn+; balanced. PDL of frozen culture is not known. Culture submitted by #0044. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Werner Syndrome - 27770

AG3594	Fibroblast, skin	30 00 00	M	WH	Skin	--	AFF BRO	546	2

The donor had symptoms of growth retardation, loss of subcutaneous fat, hyperkeratosis of palms and soles, cutis marmorata with ulcerations, lank face, baldness, high-pitched voice, bilateral cataracts, diabetes, and hypercholesterolemia (Fredrickson type IV). The family history is positive with an affected brother (AG3593A). Unaffected parents are first cousins. The cell morphology is fibroblast-like. The karyotype is 46,XY,inv(3)(q23q29)t(3;13)(p21;q32),inv(9)(p11q34); balanced. PDL of frozen culture is not known. Culture submitted by #0044. Culture medium: EEMEM 20U.

AG4110	Fibroblast, skin	13 02 00	F	WH	Skin	--	PROBAND	678	1

The donor had signs of premature aging including growth retardation, atrophic skin changes, beak-like nose, high-pitched voice, hypoplasia of clavicles, poor dentition and sparse hair. She is diagnosed with severe Werner syndrome or possibly mild progeria. The family history is negative for both syndromes. The culture was initiated on 5/24/80 using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 7. Biopsy submitted by #0126. Culture medium: EEMEM 20U.

AG7896	Lymphoblast	56 05 03	F	WH	Blood	--	PROBAND	1081	1

The affected donor had symptoms of short stature, thin skin, flat feet, increased serum triglycerides, baldness and juvenile cataracts. Her unaffected parents are first cousins, & the family history is negative. The culture was initiated on 9/18/84 and grows in suspension. The cell morphology is spherical. Culture submitted by #0309. Culture medium: R1640 20I.

AG7897	Lymphoblast	90 00 00	M	WH	Blood	+-(D)	FATHER	1081	2

The donor is clinically normal. The culture was initiated on 9/17/84 and grows in suspension. The cell morphology is spherical. Culture submitted by #0309. Culture medium: R1640 20I.

AG7898	Lymphoblast	60 00 00	F	WH	Blood	+?	SIS	1081	3

The donor is clinically normal. The culture was initiated on 9/12/84 and grows in suspension. The cell morphology is spherical. Culture submitted by #0309. Culture medium: R1640 20I.

SV-40 Virus-Transformed Werner Syndrome

AG7066B	Fibroblast, skin	29 00 00	F	CR	Skin	--	PROBAND		

AG7066B is an SV40 virus-transformed permanent line PSV 811. The culture was initiated by infection of a skin fibroblast culture from a Werner syndrome patient at PDL 3 with SV40 virus strain 777. The culture entered crisis at PDL 56 and is frozen at approximately 84 population doublings after crisis. The morphology is epithelial-like. The karyotype is abnormal with chromosome number broadly distributed in diploid range. Culture submitted by #0382. Culture medium: EEMEM 15U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Xeroderma Pigmentosum: Complementation Group A - 27870

*AG3964	Fibroblast, skin	35 00 00	M	WH	Arm	--	PROBAND		

The Egyptian donor had squamous cell carcinoma of skin. His mental and neurological condition was normal. His offspring are clinically unaffected. The biopsy was taken ante-mortem on 3/19/80. Culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male with 27% of examined cells showing chromosomal abnormalities. Complementation group assignment and DNA repair deficiency was verified using this culture (XP13CA). Culture frozen at PDL 6. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

AG6929	Fibroblast, skin	18 00 00	F	WH	Arm	--	PROBAND		

The Egyptian donor had symptoms of freckled and pigmented skin on face, keratosis on right cheek and carcinoma on nose. Family history is negative, but parents are first cousins. The culture was initiated on 7/13/83 using explants of minced skin tissue from a biopsy taken earlier and stored frozen. The cell morphology is fibroblast-like. Complementation group assignment & DNA repair deficiency were verified using this culture (XP23CA). Culture frozen at PDL 6. Biopsy submitted by #0044. Culture medium: EEMEM 20U.

AG6971	Fibroblast, skin	02 00 03	M		Arm	--	PROBAND		

The donor shows mild skin symptoms and definite neurological developmental delay. The family history is negative. The biopsy was taken ante-mortem on 4/26/83 from skin on the forearm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The complementation group assignment was done on this culture prior to submission to repository. The culture is frozen at estimated PDL 12, & was submitted by #0483. Culture medium: EEMEM 20U.

Xeroderma Pigmentosum: Complementation Group C - 27872

*AG3962	Fibroblast, skin	16 00 00	F	WH	Arm	--	PROBAND		

The Egyptian donor showed mild skin symptoms of XP, mainly depigmentation of face and hands. Her mental and neurological status was normal. The skin biopsy was taken ante-mortem on 3/19/80. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Karyotype is 46,XX; normal diploid female. Complementation group assignment & DNA repair deficiency were verified using this culture (XP11CA). Culture frozen at PDL 5. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

*AG3963	Fibroblast, skin	21 00 00	M	WH	Arm	--	PROBAND		

The Egyptian donor showed mild skin symptoms of XP, mainly depigmentation, and his mental and neurological status was normal. The skin biopsy was taken ante-mortem on 3/19/80. Culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY/46,XY,+7; 98%/2%. Complementation group assignment and DNA repair deficiency were verified using the culture (XP12CA). Culture frozen at PDL 6. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Xeroderma Pigmentosum: Complementation Group C - 27872

*AG3965	Fibroblast, skin	05 00 00	F	WH	Arm	--	PROBAND		

The Egyptian donor had severe skin symptoms of XP and a tumor of the tongue. Her mental and neurological status was normal. Her parents are first cousins. The skin biopsy was taken ante-mortem on 3/19/80. Culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Complementation group assignment & DNA repair deficiency verified using this culture (XP14CA). Culture frozen at PDL 6. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878

*AG1951	Fibroblast, skin	40 00 00	M	WH	Skin	--	PROBAND		

The donor (from USSR) had mild but typical skin defects of XP and normal mental status. The cell morphology is fibroblast-like. The karyotype is 46,XY,t(17;20)(17pter>17q25::20q13i>20qter;20pter>20q13i::17q25>17qter). The culture is frozen at passage 12 and PDL is unknown. Culture (line LHC638) submitted by #0201. Culture medium: EEMEM 20U.

AG1952	Fibroblast, skin	07 00 00	M	WH	Skin	--	PROBAND		

The donor (from USSR) showed skin defects and oligophrenia, typical for DeSanctis-Cacchione syndrome. The cell morphology is fibroblast-like. Culture is frozen at passage 12 and PDL is unknown. Culture (line LHC484) submitted by #0201. Culture medium: EEMEM 20U.

AG4316A	Fibroblast, skin	15 00 00	M	WH	Skin	--	PROBAND		

The Egyptian donor had severe skin symptoms of XP, but his mental and neurological status is normal. The biopsy was taken ante-mortem on 3/19/80. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Repair deficiency was determined in this culture (XP17CA) prior to submission to Repository. The culture is frozen at an estimated PDL 8 and was submitted by #0023. Culture medium: EEMEM 20U.

AG2872	Fibroblast, skin	24 00 00	F	WH	Lower back	--	PROBAND	318	1

The biopsy was taken ante-mortem on 1/19/78 from a Chilean donor. The culture (XP1SA) was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

AG2871	Fibroblast, skin	25 00 00	F	WH	Lower back	+?	SIS	318	2

The Chilean donor is clinically unaffected and is a possible heterozygote for XP. The biopsy was taken ante-mortem on 1/19/78. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878

AG7962 Lymphoblast — 24 00 00 — F — WH — Blood — -- — PROBAND — 1086 — 1

The donor (Finnish) is clinically affected with symptoms of photophobia, freckling, skin atrophy on face, abnormal hearing and speech, moderate impairment of intellectual functions, and ataxia. The family history is positive with two similarly affected sibs. The blood specimen was obtained on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension, & the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7963 Fibroblast, skin — 24 00 00 — F — WH — Arm — -- — PROBAND — 1086 — 1

The donor (Finnish) is clinically affected with symptoms of photophobia, freckling, skin atrophy on face, abnormal hearing and speech, moderate impairment of intellectual functions, and ataxia. The family history is positive with two similarly affected sibs. The skin biopsy was taken ante-mortem 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The culture is frozen at PDL 4. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

AG7952A Lymphoblast — 68 00 00 — M — WH — Blood — +-(0) — FATHER — 1086 — 2

The donor (Finnish) is clinically normal. The blood specimen was obtained on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7953 Fibroblast, skin — 68 00 00 — M — WH — Arm — +-(0) — FATHER — 1086 — 2

The donor (Finnish) is clinically normal. The skin biopsy was obtained ante-mortem on 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

AG7954A Lymphoblast — 16 00 00 — M — WH — Blood — -- — AFF BRO — 1086 — 3

The donor (Finnish) is clinically affected with symptoms of photophobia, freckling, atrophy & telangiectasia of facial skin, severe neurological abnormalities with low IQ, and growth deficiency. The family history is positive with two similarly affected sibs. The blood specimen was obtained on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension & the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7955 Fibroblast, skin — 16 00 00 — M — WH — Arm — -- — AFF BRO — 1086 — 3

The donor (Finnish) is clinically affected with symptoms of photophobia, freckling, atrophy and telangiectasia of skin on face, severe neurological abnormalities with low IQ and growth deficiency. The family history is positive with two similarly affected sibs. The skin biopsy was taken ante-mortem on 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 3. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878

AG7956	Lymphoblast	31 00 00	F	WH	Blood	+?	SIS	1086	4

The donor (Finnish) is clinically normal. The blood specimen was obtained on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7957	Fibroblast, skin	31 00 00	F	WH	Arm	+?	SIS	1086	4

The donor (Finnish) is clinically normal. The skin biopsy was taken ante-mortem on 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

AG7958	Lymphoblast	29 00 00	F	WH	Blood	+?	SIS	1086	5

The donor (Finnish) is clinically normal. The blood specimen was obtained on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7959	Fibroblast, skin	29 00 00	F	WH	Arm	+?	SIS	1086	5

The donor (Finnish) is clinically normal. The skin biopsy was taken ante-mortem on 11/30/84 from the inner aspect of the upper arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

AG7960	Lymphoblast	20 00 00	F	WH	Blood	+?	SIS	1086	6

The donor (Finnish) is clinically normal. The blood specimen was taken on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7961	Fibroblast, skin	20 00 00	F	WH	Arm	+?	SIS	1086	6

The donor (Finnish) is clinically normal. The skin biopsy was taken ante-mortem on 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

HUMAN CELL CULTURES: AGING

Xeroderma Pigmentosum: Complementation Group Undetermined - 27870 to 27878

AG7950	Lymphoblast	30 00 00	M	WH	Blood	++	BRO-IN-LAW	1086	7

The donor (Finnish) is clinically normal and is the husband of AG7958-AG7959. The blood specimen was taken on 11/30/84. The culture was initiated by transformation of lymphocytes with EB virus. The culture grows in suspension and the cell morphology is spherical. Blood specimen submitted by #0525. Culture medium: R1640 15I.

AG7951	Fibroblast, skin	30 00 00	M	WH	Arm	++	BRO-IN-LAW	1086	7

The donor (Finnish) is clinically normal and is the husband of AG7958-AG7959. The skin biopsy was taken ante-mortem on 11/30/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture frozen at PDL 6. Biopsy submitted by #0525. Culture medium: EEMEM 20U.

Xeroderma Pigmentosum: Variants - 27875

*AG3961	Fibroblast, skin	19 00 00	F	WH	Arm	--	PROBAND		

The Egyptian donor had mild skin symptoms of XP and a normal mental and neurological status. The family history includes one similarly affected sister. The biopsy was taken ante-mortem on 3/19/80. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. This culture (XP10CA) showed abnormal replication of UV-damaged DNA. Culture frozen at PDL 6. Biopsy submitted by #0023. Culture medium: EEMEM 20U.

CHROMOSOMAL ABERRATIONS: AGING

Aneuploid Chromosome Number - Non-Trisomic

AG7443	Fibroblast, skin	00 05 00	F	WH	Skin		PROBAND		

The donor was diagnosed with Turner syndrome (45,X) based on karyotype of blood cells. The culture was initiated on 2/25/84 using explants of minced skin tissue obtained post-mortem from this fetus of approximately 20 weeks gestational age. The cell morphology is fibroblast-like. The culture is frozen at PDL 6 and was submitted by #0107. Culture medium: EEMEM 20U.

AG7549	Fibroblast, skin	00 04 15	F		Skin		PROBAND		

The donor was diagnosed with Turner syndrome, and the karyotype of the culture prior to submission to the Repository was 45,X. The biopsy was taken post-mortem from this fetus of 18 weeks gestational age. The culture was initiated on 4/18/84 using explants of minced tissue. The culture is frozen in the 3rd passage at an estimated PDL 8. Culture submitted by #0107. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REPO	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

CHROMOSOMAL ABERRATIONS: AGING

Aneuploid Chromosome Number - Non-Trisomic

AG8006 Lymphoblast 42 05 07 F WH Blood PROBAND

The donor was diagnosed with Turner's syndrome based on a 45,X karyotype in blood lymphocytes and from characteristic physical signs. The culture was initiated on 1/30/85 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0529. Culture medium: R1640 15I.

Aneuploid Chromosome Number - Trisomy

AG7096 Fibroblast, skin 00 05 08 M WH Thoracic skin PROBAND

The donor had typical features of Down's syndrome (trisomy 21). The skin biopsy was taken post-mortem on 10/13/83. The culture was initiated using explants of minced skin tissue. The karyotype is 47,XY,+21,9qh+ and confirms the diagnosis. The cell morphology is fibroblast-like. Culture frozen at PDL 6. Biopsy submitted by #0074. Culture medium: EEMEM 20U.

*AG7167 Fibroblast, skin 17 02 00 F BL Arm PROBAND

The donor was diagnosed with trisomy 18 and had delayed physical and psychomotor development since birth. She developed Wilms tumor at 11 years of age. The biopsy was taken ante-mortem from skin of the deltoid area. The culture was initiated on 10/20/83 using explants of minced skin tissue. The cell morphology is fibroblast-like. Karyotype is 47,XX,+18. The culture is frozen at PDL 5 and was submitted by #0322. Culture medium: EEMEM 20U.

AG7409 Fibroblast, skin 01 00 00 F WH Thoracic skin PROBAND

The donor was diagnosed with Down's syndrome (trisomy 21). The skin biopsy was taken post-mortem on 3/9/84. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 47,XX,+21 and confirms the diagnosis. Culture frozen at PDL 5. Biopsy submitted by #0107. Culture medium: EEMEM 20U.

AG4823 Fibroblast, skin 05 00 00 M Thoracic skin PROBAND

The donor had typical features of Down's syndrome (trisomy 21). The skin biopsy was taken ante-mortem. The culture was initiated on 2/10/81 using explants of minced skin tissue. The karyotype is 47,XY,+21 and confirms the diagnosis. The cell morphology is fibroblast-like. The culture was frozen at PDL 6 & was submitted by #0107. Culture medium: EEMEM 20U.

AG5024 Fibroblast, skin 01 00 00 F Thoracic skin PROBAND

The donor had typical features of Down's syndrome (trisomy 21). The skin biopsy was taken post-mortem on 6/16/81. The culture was initiated using explants of minced skin tissue. The karyotype is 47,XX,+21 and confirms the diagnosis. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0107. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REPO	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAMO	FAMNO
		CHROMOSOMAL ABERRATIONS: AGING							
	Aneuploid Chromosome Number - Trisomy								
AG5397	Fibroblast, skin	13 00 00	M	WH	Thoracic skin		PROBAND		
	The donor had typical features of Down's syndrome (trisomy 21). The skin biopsy was taken ante-mortem on 2/17/82. The culture was initiated using explants of minced skin tissue. The karyotype is 47,XY,+21 and confirms the diagnosis. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0107. Culture medium: EEMEM 20U.								
AG6872	Fibroblast, skin	01 04 25	F	WH	Thorax-abdomen		PROBAND		
	The donor had typical features of Down's syndrome (trisomy 21). The skin biopsy was taken post-mortem on 5/19/83. The culture was initiated using explants of minced skin tissue. The karyotype is 47,XX,+21 and confirms the diagnosis. The cell morphology is fibroblast-like. Culture frozen at PDL 4. Biopsy submitted by #0074. Culture medium: EEMEM 20U.								
AG6922	Fibroblast, skin	01 08 00	M	WH	Thorax-abdomen		PROBAND		
	The donor had typical features of Down's syndrome. The skin biopsy was taken post mortem on 6/23/83. The culture was initiated using explants of minced skin tissue. The karyotype is 46,XY,+21 and confirms the diagnosis. Culture frozen at PDL 3. Biopsy submitted by #0074. Culture medium: EEMEM 20U.								
AG7438A	Fibroblast, skin	00 09 00	M	BL	Thoracic skin		PROBAND		
	The donor was diagnosed with Down's syndrome (trisomy 21) and displayed typical facies. The skin biopsy was taken post-mortem on 3/15/84 and frozen. The culture was initiated on 6/7/84 using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 47,XY,+21 and confirms the diagnosis. Culture frozen at PDL 4. Biopsy submitted by #0107. Culture medium: EEMEM 20U.								
AG5120	Fibroblast	00 05 15	F		Umbilical cord		PROBAND	817	1
	This culture is derived from cord tissue obtained from a therapeutically aborted fetus of 22 weeks gestational age. The karyotype of the culture is 47,XX,+21 and confirms the diagnosis of Down's syndrome (trisomy 21). The cell morphology is fibroblast-like. The culture is frozen at PDL 3 and was submitted by #0107. Culture medium: EEMEM 20U.								
AG5121	Fibroblast	00 05 15	F		Amniotic fluid		PROBAND	817	1
	This culture is derived from the amniotic fluid obtained from a therapeutically aborted fetus of a 22 weeks gestational age. The karyotype is 47,XX,+21 and the diagnosis is Down's syndrome (trisomy 21). The culture is frozen at PDL 5 and was submitted by #0107. Culture medium: EEMEM 20U.								

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

CHROMOSOMAL ABERRATIONS: AGING

Triploid Chromosome Number

AG5025 Fibroblast, skin — 00 00 01 — A — WH — Skin — PROBAND

The donor was born prematurely with multiple congenital abnormalities typical for triploidy. The culture was initiated on 5/20/81 using explants of minced skin tissue obtained post-mortem. The karyotype is 69,XXY and confirms diagnosis. The cell morphology is fibroblast-like. The culture is frozen at PDL 5 and was submitted by #0107. Culture medium: EEMEM 20U.

AG6266 Fibroblast, skin — 00 00 02 — M — WH — Lower back — PROBAND

The donor was born with multiple congenital abnormalities typical for triploidy. The culture was initiated on 1/21/83 using explants of minced skin tissue. The karyotype is 69,XXY and confirms diagnosis. The cell morphology is fibroblast-like. Culture frozen at PDL 5. Biopsy submitted by #0012. Culture medium: EEMEM 20U.

CANCER PATIENTS: INVOLVED AND UNINVOLVED TISSUES

Acute Myelogenous Leukemia

***AG2655** Fibroblast, skin — 25 08 26 — F — WH — Skin — PROBAND — 308 — 1

The donor had developed acute myelogenous leukemia at age 10 years and is a member of a high cancer incidence family, National Cancer Institute Family 375. The family history includes three similarly affected sibs. The skin biopsy was taken on 8/28/77, fifteen years after treatment for the disease. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

***AG2648** Lymphoblast — 17 09 08 — M — WH — Blood — BRO — 308 — 2

The donor is a clinically unaffected member of a high cancer incidence family, National Cancer Family 375. The culture was initiated on 8/29/77 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Cultures from a twin brother are AG2646 and AG2647. Blood specimen submitted by #0195. Culture medium: R1640 20I.

***AG2649** Fibroblast, skin — 17 09 08 — M — WH — Skin — BRO — 308 — 2

The donor is a clinically unaffected member of a high cancer incidence family. The skin biopsy was taken ante-mortem on 8/28/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

CANCER PATIENTS: INVOLVED AND UNINVOLVED TISSUES

Acute Myelogenous Leukemia

*AG2646	Lymphoblast	17 09 08	M	WH	Blood		BRO	308	3

The donor is a clinically unaffected member of a high cancer incidence family, National Cancer Institute Family 375. The culture was initiated on 8/29/77 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Cultures from a twin brother are AG2648 and AG2649. Blood specimen submitted by #0195. Culture medium: R1640 20U.

*AG2647	Fibroblast, skin	17 09 08	M	WH	Skin		BRO	308	3

The donor is a clinically unaffected member of a high cancer incidence family. The skin biopsy was taken ante-mortem on 8/28/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 4. Cultures from a twin brother are AG2648 and AG2649. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

*AG2653	Fibroblast, skin	42 02 09	F	WH	Skin		MOTHER	308	4

The donor is unaffected for leukemia, but was treated with radium for uterine (cervical) carcinoma at age 27 years. She is a member of a high cancer incidence family, National Cancer Institute Family 375. The skin biopsy was taken ante-mortem on 8/29/77. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

*AG3607	Fibroblast, skin	44 02 01	F	WH	Skin		MOTHER	308	4

The donor is unaffected for leukemia, but was treated with radium for uterine (cervical) cancer at age 27 years and also developed rectal cancer. She is a member of a high cancer incidence family, National Cancer Institute Family 375. The skin biopsy was taken ante-mortem on 8/20/79. The culture was initiated using explants of minced skin tissue. The culture is a mosiac with karyotype 46,XX/46,XX,t(12;13)(12qter>12p11::13q34>13qter;13pter>13q34::12p11>12pter), balanced; 56%/44%. The cell morphology is fibroblast-like. A culture initiated from a biopsy taken 2 years earlier is AG2653. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

*AG2650	Lymphoblast	46 01 09	M	WH	Blood		FATHER	308	5

The donor is clinically unaffected and married to a member of a high cancer incidence family, National Cancer Institute Family 375. The culture was initiated on 8/29/77 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0195. Culture medium: R1640 20I.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

CANCER PATIENTS: INVOLVED AND UNINVOLVED TISSUES

Acute Myelogenous Leukemia

*AG2651	Fibroblast, skin	46 01 09	M	WH	Skin		FATHER	308	5

The donor is clinically unaffected and married to a member of a high cancer incidence family, National Cancer Institute Family 375. The skin biopsy was taken ante-mortem on 8/28/77. The culture was initiated using explants of minced skin tissue. The karyotype is abnormal: 46,XY,t(5;8)(5qter>5p11::8q11>8qter;8pter>8q11::5p11>5pter); balanced. Culture frozen at PDL 6. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

*AG3778	Fibroblast, skin	50 10 28	F	WH	Skin		AUNT	308	6

The donor is clinically unaffected for leukemia, but developed breast cancer (infiltrating ductal carcinoma). She is a member of a high cancer incidence family, National Cancer Institute Family 375. The skin biopsy was taken ante-mortem on 1/3/80. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 5. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

Basal Cell Nevus Syndrome (Odontogenic Keratocysts & Skeletal Anom.) - 10940

*AG3499	Fibroblast, skin	11 00 00	F	WH	Eyelid	+-	PROBAND		

The donor developed numerous basal cell carcinomas, a brain meduloblastoma (sarcoma) & an ovarian tumor by age 5 years. The family history is positive for basal cell nevus syndrome and includes an affected mother, sister & grandfather. The pedigree shows autosomal dominant inheritance. The family history is also positive for Charcot-Marie-Tooth disease. The culture was initiated on 5/9/79. The karyotype is 46,XX/46,X,t(X;5)(Xpter>Xq11::5q35>5qter;5pter>5q35::Xq11>Xqter);85%/15%. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

Multiple Hamartoma Syndrome (Cowden Disease) - 15835

*AG5328	Fibroblast, skin	02 00 00	M	WH	Arm	+-	PROBAND		

The donor had the characteristic mucocutaneous findings of Cowden disease including a cobblestone appearance of labial, buccal and gingival mucosa and verrucous lesions of the face and hands. He had a history of basal and squamous cell carcinoma and lipomas. Similarly affected family members include a brother, two sons & eight nephews. The biopsy was taken ante-mortem on 12/17/81 from skin of the inner aspect of the upper arm. Culture was initiated using explants of minced skin. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.

AG1966	Fibroblast, skin	58 00 21	F	WH	Arm	+-	PROBAND	181	1

The donor had a history of benign tumors including adenomas of the thyroid, leiomyomata of the uterus, an ovarian cyst, and papillomas of the hand. The family history shows many relatives with multiple benign tumors similar to donor's. The diagnosis is tentatively Cowden disease. The skin biopsy was taken ante-mortem on 10/10/76. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female with 40% of examined cells showing a chromatid break. Culture frozen at PDL 5. Biopsy submitted by #0195.

Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#
					CANCER PATIENTS: INVOLVED AND UNINVOLVED TISSUES				
	Multiple Hamartoma Syndrome (Cowden Disease) - 15835								
AG1964	Fibroblast, skin	62 06 03	M	WH	Arm	++	PAT 1ST COU	181	2
	The donor is clinically unaffected. The skin biopsy was taken ante-mortem on 10/10/76. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Karyotype is 46,XY,9qh+; diploid male. Culture frozen at PDL 5. Biopsy submitted by #0195. Culture medium: EEMEM 20U.								
AG1965	Fibroblast, skin	60 06 21	F	WH	Arm	++	MAT 1ST COU	181	3
	The donor is a clinically unaffected member of a cancer prone family. The skin biopsy was taken ante-mortem on 10/10/76 The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 5. Biopsy submitted by #0195. Culture medium: EEMEM 20U.								
AG1967	Fibroblast, skin		M	WH	Arm	++	PAT 1ST COU	181	4
	The donor is clinically unaffected. The skin biopsy was taken ante-mortem on 10/10/76. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XY; normal diploid male. Culture frozen at PDL 5. Biopsy submitted by #0195. Culture medium: EEMEM 20U.								
AG1968	Fibroblast, skin	49 06 29	F	WH	Arm	++	PAT 1ST COU	181	5
	The donor is clinically unaffected with Cowden disease but had developed a uterine cyst and a throat tumor. The skin biopsy was taken ante-mortem on 10/10/76. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 5. Biopsy submitted by #0195. Culture medium: EEMEM 20U.								
AG1969	Fibroblast, skin		F	WH	Arm	+-	AFF NIECE	181	6
	The donor developed papillomas of the hand and is tentatively diagnosed with Cowden disease. She developed a grade III-IV occipital astrocytoma after the skin biopsy for this culture was taken. Her mother is diagnosed with Cowden disease, and her family history is positive for benign and malignant tumors. The skin biopsy was taken ante-mortem on 10/10/76. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture frozen at PDL 4. Biopsy submitted by #0195. Culture medium: EEMEM 20U.								
	Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530								
AG4243	Lymphoblast	28 00 00	F	WH	Blood	+-	PROBAND	670	1
	The donor had developed two breast and two leg cysts, abdominal wall desmoid tumors, and multiple adenomatous polyps and carcinoma of the colon. The culture was initiated on 7/16/80 by transformation of lymphocytes with EB virus. The culture grows in suspension, and the cell morphology is spherical. Blood specimen submitted by #0384. Culture medium: R1640 20I.								

AGING CELL REPOSITORY CULTURES

HUMAN CELL CULTURES FROM PATIENTS WITH DISEASES RELEVANT TO AGING

REP#	CELL TYPE	AGE YR MO DA	SEX	RACE	ORIGIN	GENETIC STATUS	REL TO PROBAND	FAM#	FAMM#

CANCER PATIENTS: INVOLVED AND UNINVOLVED TISSUES

Polyposis, Intestinal: Type III (Gardner Syndrome) - 17530

AG4245A	Fibroblast, skin	28 00 00	F	WH	Keloid	+-	PROBAND	670	1

The donor had developed two breast and two leg cysts, abdominal wall desmoid tumors, and multiple adenomatous polyps and carcinoma of the colon. The biopsy was taken from a keloid formation on the back which formed after a previous biopsy procedure. The culture was initiated using explants of the minced keloid tissue. The cell morphology is fibroblast-like and the culture is slow growing. Culture frozen at PDL 5. Biopsy submitted by #0384. Culture medium: EEMEM 20U.

*AG4248B	Fibroblast	28 00 00	F	WH	Tumor	+-	PROBAND	670	1

The donor had developed two breast and two leg cysts, abdominal wall desmoid tumors, and multiple adenomatous polyps and carcinoma of the colon. The culture was initiated using explants of an abdominal desmoid tumor section. Culture frozen at PDL 6. Biopsy submitted by #0384. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: ARTIODACTYLA					
		GENUS: Bos					
AG2791A	taurus/cattle	Endothelial, arterial	Artery, pulmonary	00 01 00	M	8	

The culture was initiated on 10/25/77. Cells of the lumen surface of the pulmonary artery were removed by collagenase digestion and provided the primary culture material. The artery was removed from a freshly slaughtered calf. The cell morphology is polygonal. Culture submitted by #0281. Culture medium: EEMEM 20U.

*AG4762	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 04 15	M	38	90

(Holstein breed) The culture was initiated on 4/25/79 using cells released by collagenase digestion from the lumen of a thoracic aorta segment obtained from a fetal calf of approximately 18 weeks gestational age. The culture was passed several times in 5% plasma-derived serum to prevent overgrowth by smooth muscle cells. The cell morphology is polygonal. The culture tested negative for presence of bovine viruses BVD, PI-3 and BHV-1. Culture submitted by #0163. An earlier passage culture of this line is AG7680. Culture medium: EEMEM 20U.

AG7680	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 04 15	M	19	90

(Holstein breed) The thoracic aorta was obtained on 4/25/79 from a 4 to 5 month old fetus. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. Plasma-derived serum was used in the primary culture to select against possible smooth muscle contaminants. The cell morphology is polygonal. The karyotype is 60,XY; normal diploid male. The culture tested positive for Factor VIII-related antigen and angiotensin-I converting enzyme and tested negative for viruses BVD, PI-3 and BHV-1 prior to submission to the Repository. Culture submitted as line BFAE-39 by #0508. A later passage of this line is AG4762. Culture medium: HF12 20U.

AG7681	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 04 15	M	49	84

(Holstein breed) This culture is a clonal derivative of AG7680 obtained by plating single cells at CPD 10. The parental line, AG7680, from a 4 to 5 month old fetus was initiated on 4/25/79, using cells released by collagenase digestion of the lumen of the thoracic aorta. The karyotype of the parental line is 60,XY; normal diploid male. The cell morphology is polygonal. This culture tested positive for Factor VIII-related antigen and angiotensin-I converting enzyme, and negative for viruses BVD, PI-3 & BHV-1 prior to submission to the Repository. Culture submitted as line BFAE-39b by #0508. Culture medium: HF12 20U.

AG7682	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 04 15	M	50	96

(Holstein breed) This culture is a clonal derivative of AG7680 obtained by plating single cells at CPD 10. The parental line, AG7680, from a 4 to 5 month old fetus was initiated on 4/25/79, using cells released by collagenase digestion of the lumen of the thoracic aorta. The karyotype is 60,XY; normal diploid male. The cell morphology is polygonal. This culture tested positive for Factor VIII-related antigen and angiotensin-I converting enzyme, & negative for viruses BVD, PI-3 and BHV-I prior to submission to the Repository. Culture submitted as line BFAE-39e by #0508. Culture medium: HF12 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: ARTIODACTYLA					
		GENUS: Bos					
AG7680	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 04 15	M	48	75

(Holstein breed) This culture is a clonal derivative of AG7680 obtained by plating single cells at CPD 10. The parental line, AG7680, from a 4 to 5 month old fetus was initiated on 4/25/79, using cells released by collagenase digestion of the lumen of the thoracic aorta. The karyotype of the parental line is 60,XY; normal diploid male. The cell morphology is polygonal. This culture tested positive for Factor VIII-related antigen and angiotensin-I converting enzyme, and negative for viruses BVD, PI-3 & BHV-1 prior to submission to the Repository. Culture submitted as line BFAE-39k by #0508. Culture medium: HF12 20U.

AG7684	taurus/cattle	Endothelial, arterial	Aorta, thoracic	00 05 00	F	16	110

(Holstein breed) The thoracic aorta was obtained on 5/17/79 from a 5 month old fetus. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. Plasma-derived serum was used in the primary culture to select against possible smooth muscle contaminants. The cell morphology is polygonal. Culture submitted as line BFAE-45 by #0508. Culture medium: HF12 20U.

AG8132	taurus/cattle	Endothelial, arterial	Aorta, thoracic	02 00 00	F	11	

(Holstein breed) The thoracic aorta was removed 4 hours post-mortem on 4/18/85. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. The cell morphology is polygonal. Culture submitted by #0477. The culture was grown on gelatin-coated flasks, and the medium was supplemented with crude ECGS (0.24mg/ml) and heparin (0.1mg/ml). Culture medium: AMMC 20U (with supplements).

AG4907	taurus/cattle	Mesothelial	Lung		F	31	40

(Holstein breed) The culture was initiated from the lung of a fetal calf on 12/27/79. The pleura was mechanically stripped from the lung, digested with trypsin, and released cells were plated. The cell morphology is epithelial-like. The karyotype is 60,XX; normal diploid female. Culture submitted by #0388. Culture medium: EEMEM 20U.

AG4388	taurus/cattle	Smooth muscle, vascular	Aorta, thoracic	00 03 00	M	28	

The tissue of origin for this culture is the medial layer of the upper thoracic aorta. The culture was initiated 9/80. The cell morphology is fibroblast-like. The karyotype is 60,XY; normal diploid male. The cells have a strong histamine signal (receptor) and deposit collagen post-confluence with addition of ascorbate. Culture submitted by #0375. Culture medium: EEMEM 20U.

		GENUS: Equus					
AG7979	caballus/horse	Endothelial, arterial	Aorta, thoracic	00 08 00	M	17	

(Standardbred horse) The aorta was removed immediately post-mortem on 12/7/84. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. The cell morphology is polygonal, and 'sprouter' cells appear at confluence. The cells were grown on gelatin-coated flasks, and the medium was supplemented with crude ECGS (0.24mg/ml) and heparin (0.1mg/ml). Culture submitted by #0477. Culture medium: AMMC 20U (with supplements). Other cultures derived from this horse are AG7980 and AG7984.

NONHUMAN CELL CULTURES

REPO	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	POL FROZEN	IN VITRO LIFE-SPAN
		ORDER: ARTIODACTYLA					
		GENUS: Equus					
AG8030	caballus/horse	Endothelial, arterial	Aorta, thoracic	02 00 00	F	11	

(Standardbred horse) The thoracic aorta was removed 1 hour post-mortem on 2/27/85. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. The cell morphology is polygonal. Culture submitted by #0477. The culture was grown on gelatin-coated flasks, and the medium was supplemented with crude ECGS (0.24mg/ml) and heparin (0.1mg/ml). Culture medium: AMMC 20U (with supplements). Other cultures derived from this horse are AG8029 and AG8031.

AG7984	caballus/horse	Fibroblast, skin	Thigh	00 08 00	M	5	

(Standardbred horse) The skin biopsy was taken 1 hour post-mortem on 12/7/84 from the inner thigh. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture submitted by #0477. Culture medium: EEMEM 20U. Other cultures derived from this horse are AG7979 and AG7980.

AG8029	caballus/horse	Fibroblast, skin	Thigh	02 00 00	F	2	

(Standardbred horse) The skin biopsy was taken post-mortem on 2/27/85 from the inner thigh. The culture was initiated using explants of minced skin. The cell morphology is fibroblast-like. Biopsy obtained by #0477. Culture medium: EEMEM 20U. Other cultures derived from this horse are AG8030 and AG8031.

AG7906	caballus/horse	Fibroblast, skin	Thigh	03 00 00	F	3	

(Thoroughbred horse) The skin biopsy was taken immediately post-mortem on 11/5/84 from the right inner thigh. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Culture submitted by #0477. Culture medium: EEMEM 20U. A smooth muscle culture derived from this horse is AG7937A.

AG8032	caballus/horse	Fibroblast, skin	Thigh	06 00 00	F	3	

(Standardbred horse) The skin biopsy was taken 1 hour post-mortem on 3/26/85 from the inner thigh. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Biopsy obtained by #0477. Culture medium: EEMEM 20U. A smooth muscle culture derived from this horse is AG8034.

AG7980	caballus/horse	Smooth muscle, vascular	Aorta, thoracic	00 08 00	M	14	

(Standardbred horse) The thoracic aorta was removed immediately post-mortem on 12/7/84. Strips of the medial layer were minced, treated with collagenase and then elastase. Cells released by this treatment were used for the primary culture. The cell morphology is fibroblast-like, & the cells form layered sheets at post-confluence. Culture submitted by #0477. Culture medium: EEMMC 20U. Other cultures derived from this horse are AG7979 and AG7984.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	POL FROZEN	IN VITRO LIFE-SPAN
		ORDER: ARTIODACTYLA					
		GENUS: Equus					
AG8031	caballus/horse	Smooth muscle, vascular	Aorta, thoracic	02 00 00	F	13	

(Standardbred horse) The thoracic aorta was removed 1 hour post-mortem on 2/27/85. Strips of medial layer were minced, treated with collagenase and then elastase. Cells released by this treatment were used for the primary culture. The cell morphology is fibroblast-like, and the cells form layered sheets at post-confluence. Culture submitted by #0477. Culture medium: EEMMC 20U. Other cultures derived from this horse are AG8029 and AG8030.

AG7937A	caballus/horse	Smooth muscle, vascular	Aorta, thoracic	03 00 00	F	3	

(Thoroughbred horse) The aorta was removed 2 hours post-mortem on 11/5/84. Strips of the medial layer were removed, minced, and used as explants to initiate the culture. The cell morphology is fibroblast-like, & the cells form layered sheets at post-confluence. Culture submitted by #0477. Culture medium: EEMMC 20U. A fibroblast culture derived from this horse is AG7906.

AG8034	caballus/horse	Smooth muscle, vascular	Aorta, thoracic	06 00 00	F	15	

(Standardbred horse) The thoracic aorta was removed post-mortem on 2/26/85. Strips of the medial layer were minced and treated with collagenase then elastase. Cells released by this treatment were used for primary culture. The cell morphology is fibroblast-like, and the cells form layered sheets at post-confluence. Culture submitted by #0477. Culture medium: EEMEM 20U. A fibroblast culture from this horse is AG8032.

		GENUS: Sus					
AG8115	scrofa/pig	Endothelial, arterial	Aorta, thoracic	01 01 00	F	7	

(Duroc breed) The aorta was taken immediately post-mortem on 4/9/85. The culture was initiated using cells released from the lumen of the vessel by collagenase treatment. The cell morphology is endothelial. Culture submitted by #0477. The cells were grown on gelatin-coated flasks, and the medium was supplemented with crude ECGS (0.24mg/ml) and heparin (0.1mg/ml). Culture submitted by #0477. Culture medium: AMMC 20U (with supplements).

AG5056	scrofa/pig	Epithelial, lens	Eye		F	21	

The culture was initiated on 11/6/79 by plating a suspension of single cells obtained from the lens capsule of an adult pig of unknown age. The cell morphology is epithelial-like. The cell line is heteroploid with all examined cells being chromosomally abnormal. Culture submitted by #0400. Culture medium: EEMEM 20U.

		ORDER: CARNIVORES					
		GENUS: Canis					
AG8059	familiaris/dog	Endothelial, arterial	Aorta, thoracic	04 04 29	F	16	

(Beagle breed) The thoracic aorta was removed post-mortem on 3/5/85. The culture was initiated using cells released from the lumen of the vessel by collagenase digestion. The cell morphology is polygonal. Culture submitted by #0477. The culture was grown on gelatin-coated flasks. The medium was supplemented with crude ECGS (0.24mg/ml) & heparin (0.1mg/ml). Culture medium: AMMC 20U (with supplements).

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: CARNIVORES					
		GENUS: Canis					
AG7454	familiaris/dog	Fibroblast, skin	Abdomen	05 04 04	F	3	

(Beagle breed) The skin biopsy was taken immediately post-mortem on 3/28/85. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like, and the karyotype is 78,XX; normal diploid female. Biopsy obtained from #0336.

AG8052	familiaris/dog	Fibroblast, skin	Abdomen	07 03 23	F	3	

(Beagle breed) The skin biopsy was taken post-mortem on 3/5/85. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Biopsy obtained from #0531. Culture medium: EEMEM 20U.

		ORDER: LAGOMORPHA					
		GENUS: Oryctolagus					
*AG4676	cuniculus/rabbit	Epithelial, lens	Eye	00 00 05	F	105	

The culture was initiated on 9/2/79 from the ocular lens capsule of a New Zealand white rabbit. The cell morphology is epithelial-like. The karyotype is 44,XX; normal diploid female. Culture submitted by #0386. Culture medium: EEMEM 20U.

*AG4677	cuniculus/rabbit	Epithelial, lens	Eye	01 06 00	F	29	

The culture was initiated on 9/1/80 from the ocular lens capsule of a New Zealand White rabbit. The cell morphology is epithelial-like. The karyotype is 44,XX; normal diploid female. The culture can be carried for an additional 40 passages after recovery from liquid nitrogen storage. Culture submitted by #0386. Culture medium: EEMEM 20U.

		ORDER: PRIMATES					
		GENUS: Ateles					
AG5352	geoffroyi geoffroyi	Fibroblast, skin	Thigh	00 00 03	M	7	

This black-handed spider monkey culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 34,XY; normal diploid male. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Callicebus					
AG6115	molloch pallescens/Para. titi	Fibroblast, skin	Thigh	08 00 00	M	9	

The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The age of the donor is estimated. Culture submitted by #0408. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REPO	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: PRIMATES					
		GENUS: Erythrocebus					
AG6254	patas/patas monkey	Fibroblast, skin	Arm	20 00 00	F	4	

The biopsy was taken ante-mortem on 1/11/83 from skin of the inner aspect of the upper arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 54,XX; normal diploid female. The animal's birthdate is not known, & its age is estimated. Biopsy submitted by #0449. Culture medium: EEMEM 20U.

AG6116	patas/patas monkey	Fibroblast, skin	Thigh	00 06 00	F	8	

The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 54,XX; normal diploid female. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Gorilla					
AG5251	gorilla/lowland gorilla	Fibroblast, skin	Arm	04 00 00	F	10	

The culture was initiated from explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 48,XX,15p+; diploid female. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Lemur					
AG7100A	catta/ring-tailed lemur	Fibroblast, skin	Arm	00 03 25	F	4	

The skin biopsy was obtained 10/17/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 56,XX,inv 10q; diploid female. Biopsy submitted by #0477. Culture medium: EEMEM 20U.

AG7099A	catta/ring-tailed lemur	Fibroblast, skin	Arm	11 04 03	F	6	

The skin biopsy was obtained 10/17/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 56,XX; normal diploid female. Biopsy submitted by #0477. Culture medium: EEMEM 20U.

AG5357	fulvus/brown lemur	Fibroblast, skin	Thigh	14 00 00	M	12	

The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. The karyotype is 60,XY; diploid male, and 33% of examined cells carried translocations involving chromosome #2. Culture submitted by #0408. Culture medium: EEMEM 20U.

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	POL FROZEN	IN VITRO LIFE-SPAN
		ORDER: PRIMATES					
		GENUS: Lagothrix					
AG5356	lagotricha/Wooly monkey	Fibroblast, skin	Thigh	17 00 00	F	9	

The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. The karyotype is 62,XX; normal diploid female. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Macaca					
AG7130A	fuscata/Japanese macaque	Fibroblast, skin	Arm	00 06 09	F	3	

The skin biopsy was obtained ante-mortem on 10/31/83 from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7098A	mulatta/rhesus monkey	Fibroblast, skin	Arm	00 00 08	F	5	

The skin biopsy was obtained 10/17/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG6249	mulatta/rhesus monkey	Fibroblast, skin	Arm	00 09 22	F	3	

The skin biopsy was obtained 1/11/83, ante-mortem, from the inner aspect of the upper arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7107A	mulatta/rhesus monkey	Fibroblast, skin	Arm	02 05 19	F	5	

The skin biopsy was obtained 10/20/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7127A	mulatta/rhesus monkey	Fibroblast, skin	Arm	05 02 14	F	5	

The skin biopsy was obtained 10/31/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REPO	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: PRIMATES					
		GENUS: Macaca					
AG6252	mulatta/rhesus monkey	Fibroblast, skin	Arm	06 06 27	F	3	

The skin biopsy was obtained 1/11/83, ante-mortem, from the inner aspect of the upper arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Biopsy submitted by #0449. Culture medium: EEMEM 20U.

AG7128A	mulatta/rhesus monkey	Fibroblast, skin	Arm	11 00 01	F	5	

The skin biopsy was obtained 10/31/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7108A	mulatta/rhesus monkey	Fibroblast, skin	Arm	17 03 16	F	5	

The skin biopsy was obtained 1/11/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Biopsy submitted by #0449. Culture medium: EEMEM 20U.

AG6253	mulatta/rhesus monkey	Fibroblast, skin	Arm	18 00 00	F	4	

The age of the donor is a minimum estimation. The donor was acquired by the submitter as an adult, and the age given is the sum of the years with submitter plus the minimum age for an adult rhesus monkey. The skin biopsy was obtained on 1/11/83, ante-mortem, from the inner aspect of the upper arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Biopsy submitted by #0449. Culture medium: EEMEM 20U.

AG7109A	mulatta/rhesus monkey	Fibroblast, skin	Arm	27 00 00	F	6	

The age of the donor is a minimum estimation. The donor was acquired as an adult at the Oregon Regional Primate Center, and the age given is the sum of the years at this center plus 5 years (the minimum age for an adult rhesus monkey). The skin biopsy was obtained on 10/20/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7110A	mulatta/rhesus monkey	Fibroblast, skin	Arm	27 00 00	F	6	

The age of the donor is a minimum estimation. The donor was acquired as an adult at the Oregon Regional Primate Center, and the age given is the sum of the years at this center plus 5 years (the minimum age for an adult rhesus monkey). The skin biopsy was obtained on 10/20/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XX; normal diploid female. Culture submitted by #0477. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
	ORDER: PRIMATES						
	GENUS: Macaca						
AG7923	nemestrina/pigtailed macaque	Fibroblast, skin	Arm	15 03 18	M	4	

The skin biopsy was taken ante-mortem on 11/14/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Biopsy obtained by #0477. Culture medium: EEMEM 20U.

AG7921	nemestrina/pigtailed macaque	Fibroblast, skin	Arm	24 10 03	M	5	

The skin biopsy was taken ante-mortem on 11/14/84 from the inner aspect of the upper left arm. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. Biopsy obtained by #0477. Culture medium: EEMEM 20U.

AG7102A	nigra/Celebes ape	Fibroblast, skin	Arm	00 04 20	M	3	

The skin biopsy was obtained 10/17/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 42,XY; normal diploid male. Culture submitted by #0477. Culture medium: EEMEM 20U.

AG7101A	nigra/Celebes ape	Fibroblast, skin	Arm	19 03 04	M	2	

The skin biopsy was obtained 10/17/83, ante-mortem, from the inner aspect of the upper left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. Culture submitted by #0477. Culture medium: EEMEM 20U.

	GENUS: Pan						
AG5253	paniscus/pigmy chimpanzee	Fibroblast, skin	Thigh	08 00 00	M	11	

The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. The karyotype is 48,XY; normal diploid male. Culture submitted by #0408. Culture medium: EEMEM 20U.

AG6939A	troglodytes/chimpanzee	Fibroblast, skin	Arm	09 00 00	M	7	

The skin biopsy was taken ante-mortem on 7/25/83 from the medial surface of the left arm. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype is 48,XY; normal diploid male. Biopsy was submitted by #0325. Culture medium: EEMEM 20U.

AGING CELL REPOSITORY CULTURES

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: PRIMATES					
		GENUS: Pongo					
AG6105	pygmaeus/orangutan	Fibroblast, skin	Thoraco-lumbar junction	26 00 00	F	3	

The donor was captured in the wild at an estimated age of 6 years & thereafter reared in a zoo. Skin biopsy was taken ante-mortem on 10/12/82. The culture was initiated using explants of minced tissue. The cell morphology is fibroblast-like. The karyotype of her lymphocytes is 48,XX; normal diploid female & is homozygous for Sumatran chromosome #2 polymorphism. Biopsy submitted by #0220. Culture medium: EEMEM 20U.

AG6209	pygmaeus/orangutan	Fibroblast, skin	Thoraco-lumbar junction	27 10 05	F	2	

The skin biopsy was taken ante-mortem on 11/30/82. The culture was initiated using explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 48,XX; normal diploid female and homozygous for Sumatran chromosome #2 polymorphism. Biopsy submitted by #0220. Culture medium: EEMEM 20U.

AG5252	pygmaeus/orangutan	Fibroblast, skin	Thigh	08 00 00	M	11	

(Bornean orangutan) The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. The karyotype is 48,XY; normal diploid male. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Presbytis					
AG5307	entellus/Hanuman langur	Fibroblast, skin	Thigh	00 00 01	F	12	

The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. Culture submitted by #0408. Culture medium: EEMEM 20U.

		GENUS: Saguinus					
AG5313	fuscicollis/wht-lipped tamarin	Fibroblast, skin	Thigh	00 00 01	F	11	

The culture was initiated from explants of skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX, 3q+; diploid female. Culture submitted by #0408. Culture medium: EEMEM 20U.

AG5308	labiatus/red-bellied tamarin	Fibroblast, skin	Thigh	06 00 00	F	11	

The culture was initiated from explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 46,XX; normal diploid female. Culture submitted by #0408. Culture medium: EEMEM 20U.

NONHUMAN CELL CULTURES

REP#	SPECIES/COMMON NAME	CELL TYPE	ORIGIN	AGE YR MO DA	SEX	PDL FROZEN	IN VITRO LIFE-SPAN
		ORDER: PRIMATES					
		GENUS: Saimiri					
AG5311	sciureus/squirrel monkey	Fibroblast, skin	Thigh		F	11	

The culture was initiated from explants of minced skin tissue. The cell morphology is fibroblast-like. The karyotype is 44,XX; normal diploid female. Animal's age is not known. Culture submitted by #0408. Culture medium: EEMEM 20U.

		ORDER: RODENTIA					
		GENUS: Mus					
AG5055	musculus/mouse	Epithelial, lens	Eye	00 00 01	M	53	

Mouse strain CD-1 (Charles River outbred line). The culture was initiated on 1/29/80 using cells released by trypsin treatment of a lens capsule which previously had been maintained in organ culture for 5 days. The cells grow abnormally in culture, and the cell morphology is epithelial-like. This line is heteroploid with all examined cells being chromosomally abnormal. Culture submitted by #0400. Culture medium: EEMEM 20U.

		GENUS: Rattus					
AG4765	norvegicus/rat	Mesothelial	Lung	00 00 10	A	33	

Culture derived from a Fisher 344 rat of Charles River strain. The culture was initiated on 2/8/78 by trypsin digestion of an intact left lung lobe. The released cells were plated on cover-slip fragments, and fragments with only epithelial cells were passaged. The cell morphology is epithelial-like. Karyotype is near diploid: 41,X/41,X,11q+; 60%/40%. This culture may have an unlimited life-span. Culture submitted by #0388. Culture medium: EEMEM 20U.

APPENDIX A

CODE FOR CULTURE MEDIA

For reference see: Morton, Helen C., A survey of Commercially Available Tissue Culture Media. In Vitro 6:89-108, 1970.

The code consists of an abbreviation of the culture medium followed by a number, indicating the percentage of fetal calf serum, and the letter I or U. I indicates that the fetal calf serum is heat inactivated at 60°C for 1/2 hour before it is added to the culture medium. U indicates that the fetal calf serum is used without prior inactivation (uninactivated). The abbreviations are as follows:

EEMEM - Minimum Essential Medium Eagle in Earle BSS with 2x concentration of essential and non-essential amino acids and vitamins

HEMEM - Minimum Essential Medium in Hank BSS, with 2x concentration of essential and nonessential amino acids and vitamins

R1640 - Roswell Park Memorial Institute Medium 1640

MC5A - McCoy 5A Medium, Iwakata and Grace Modification

DUMEM - Dulbecco Minimal Essential Medium

HF12 - Ham's F12 Medium

HF10 - Ham's F10 Medium

FRZBX - Frozen Biopsy

MES - Equal volumes of 199 Medium in Earle BSS and MCDB 202 Medium

EEMMC - Equal volumes of Minimum Essential Medium Eagle (in Earle BSS with 2x concentration of essential and non-essential amino acids and vitamins) and MCDB 104 medium

AMMC - Equal volumes of Alpha minimum essential medium Eagle (with nucleosides) and MCDB 104 medium

F12MC - Equal volumes of Ham's F12 medium and MCDB 104 medium.

APPENDIX B

SUBMITTER CODE

THE HUMAN GENETIC MUTANT CELL REPOSITORY IS INDEBTED TO THE FOLLOWING INVESTIGATORS WHO GRACIOUSLY SUBMITTED BIOPSIES, CULTURES, AND DATA FOR CHARACTERIZATION.

0001 Akstein	E	The Sheba Medical Center Cytogenetics Lab TelHashome Israel
0002 Alfi	D	Childrens Hosp, Div Med Gen 4650 Sunset Blvd LosAngeles CA 90054 USA
0003 Allderdice	P	Memorial University Dept of Medicine St Johns NF Canada
0004 Aula	P	U of Helsinki, Dep Pathology Haartmaninkatu 3 Helsinki 00290 Finland
0005 Barranco	S	U of Texas Med Sch Div of Cell Biology Galveston TX 77550 USA
0006 Beaudet	A	Baylor Col Med, Dep of Ped Rm 304E 1200 Moursund Ave Houston TX 77030 USA
0007 Benke	P	PO Box 016820 Miami FL 33101 USA
0008 Beratis	N	Med Gen Ins for Basic Res Men Ret 1050 Forest Hill Road Staten Isl NY 10314 USA
0009 Beutler	E	Scripps Cl & Res Fdn Dep Bas & Clin Res 10666 N Torrey Pines Rd La Jolla CA 92037 USA
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APPENDIX C

REFERENCES

The following pages list references which include clinical descriptions of patients and their families from whom specimens stored in the Repository were obtained as well as publications detailing investigations utilizing cultures obtained from the Repository. This list is updated annually to provide a comprehensive bibliography for each culture. This goal could be realized more effectively if investigators would identify cultures in their publications by their GM or AG numbers and forward reprints to the Repository.

The information sheets accompanying each cell shipment include the title of the article in addition to the literature citation.

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6979	Am J Hum Genet 35:101A (1983) Kukolich
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7254	Cytogenet Cell Genet 38:238 (1984) Azen
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7977	Diabetes 27:1112 (1978) Fajans

APPENDIX D

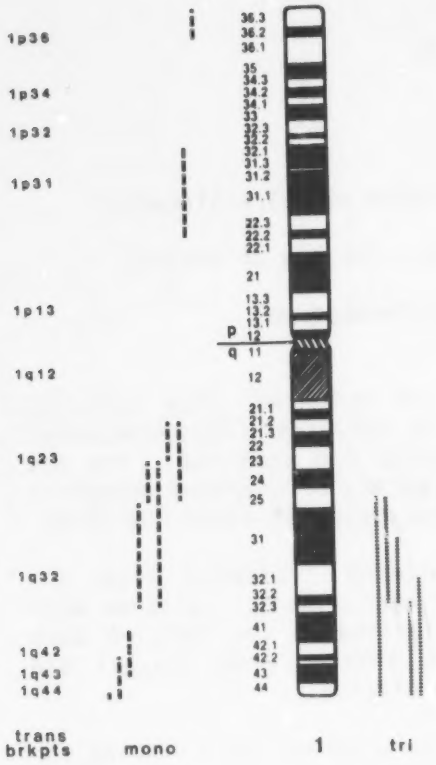
Diagrams Indicating Cultures Available with Translocation Breakpoints and Regions of Monosomy, Trisomy or Greater Dosages for Each Human Chromosome

This year, the breakpoint map of cultures with balanced chromosome translocations and the map of cultures with chromosomal monosomies or trisomies have been updated and combined. The new format uses the ISCN (1981) standard diagrams of G-banded metaphase chromosomes which represent a haploid karyotype of about 550 bands.

Translocation breakpoints for cultures available from the Repository are shown to the far left of each figure. Cultures with monosomic portions of the genome are located to the left of each diagram and cultures with trisomic or greater gene dosages are located to the right of each chromosome diagram.

Immediately following the chromosome diagrams is a listing for each of the chromosomes which details the cultures available with the specific translocation breakpoints or dosages depicted in the diagrams. Additional information for a culture of interest may be obtained by referring to the catalog text. The location of a cell culture in the catalog may be found by consulting the Repository number versus page number index (Appendix H).

All of the cultures depicted in the diagrams have been verified by karyotype by the staff of the Human Genetic Mutant Cell Repository after recovery of seed stock cultures from liquid nitrogen storage. Investigators utilizing these cultures are encouraged to inform the Repository of discrepancies between banding analysis and analyses performed using molecular biologic techniques. These additional kinds of information will contribute greatly to the quality of this unique collection.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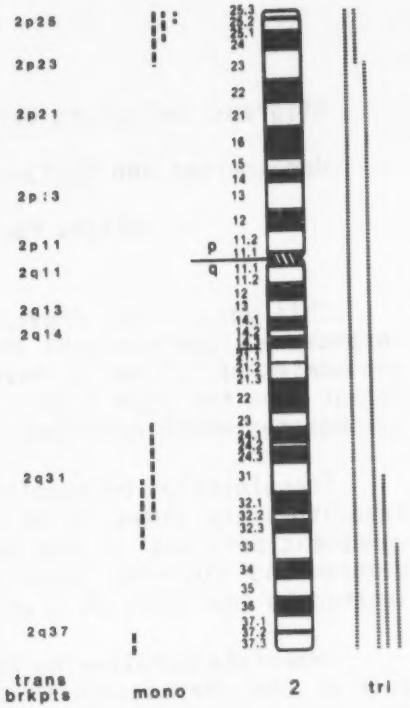
1
tri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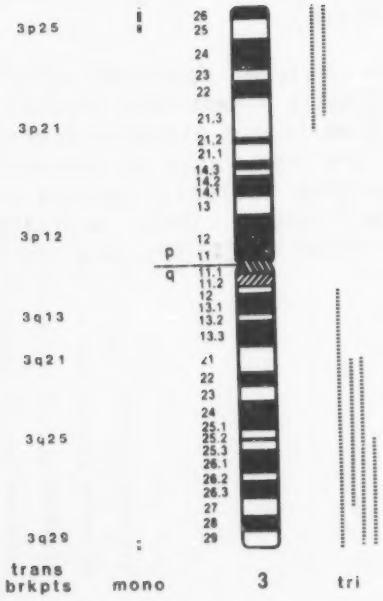
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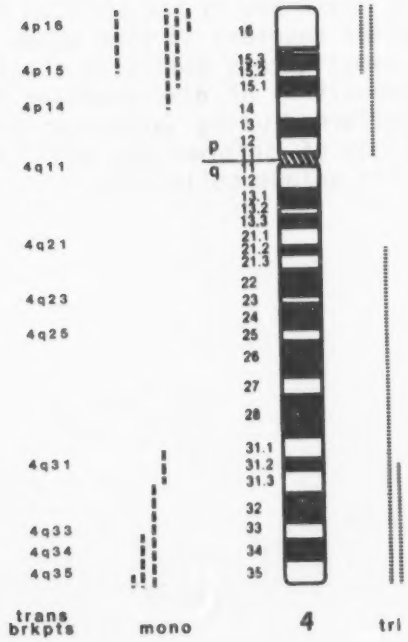
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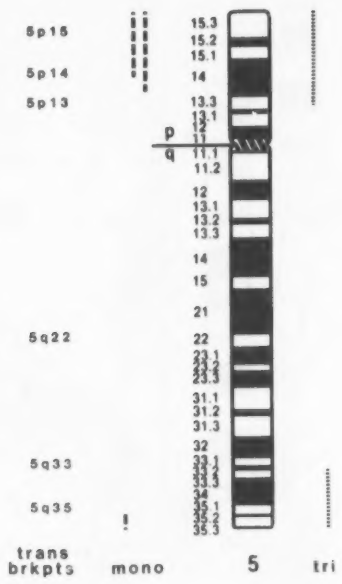
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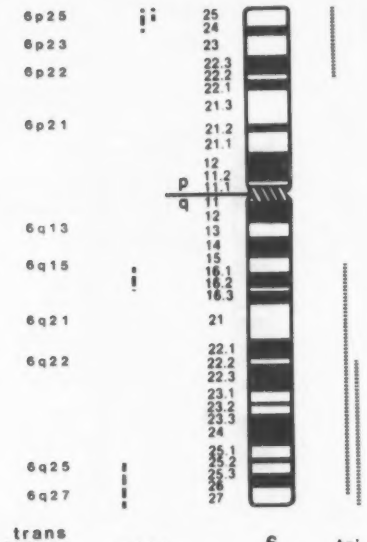
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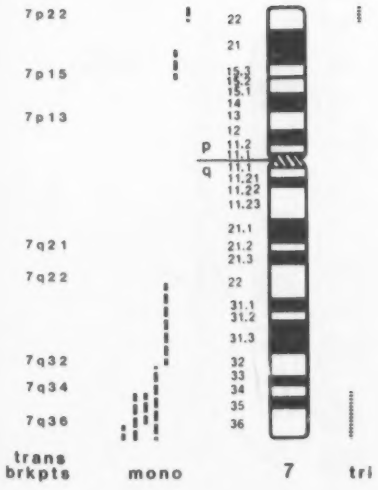
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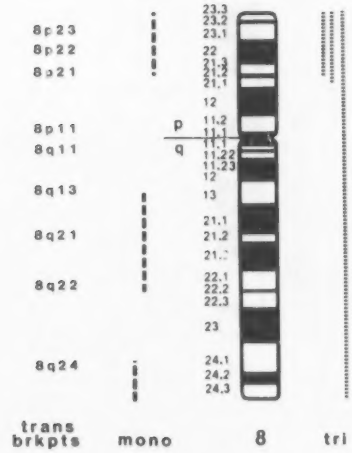
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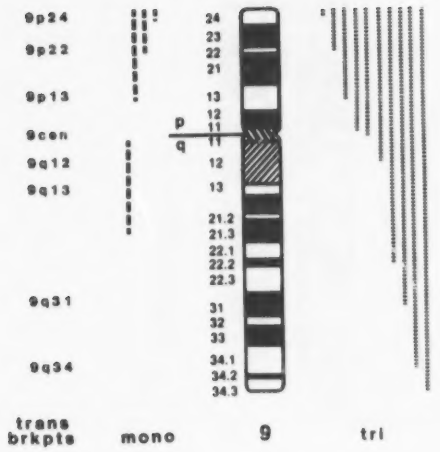
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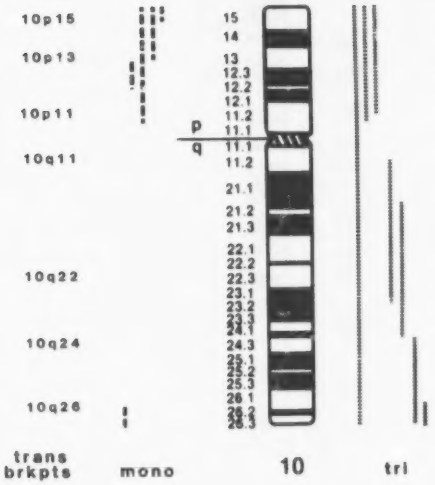
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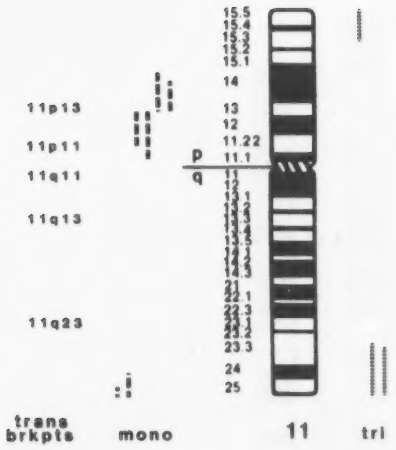
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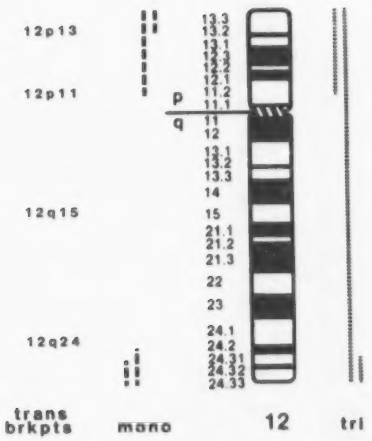
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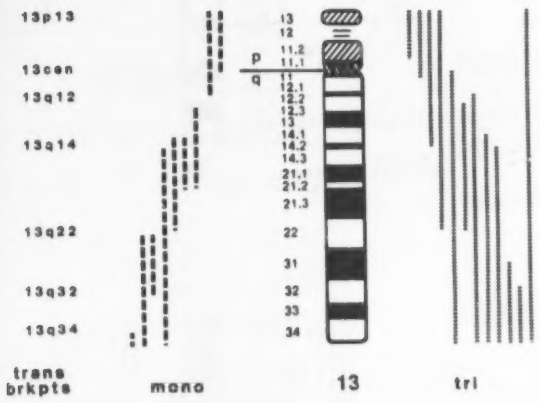
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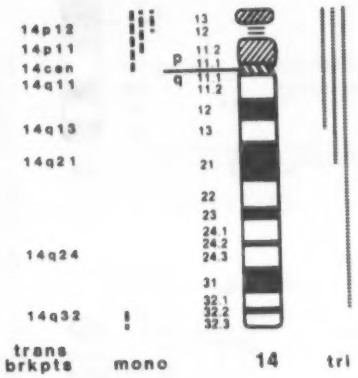
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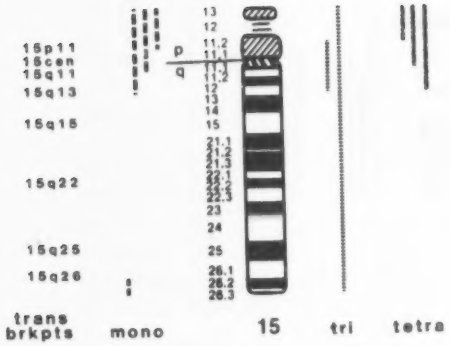
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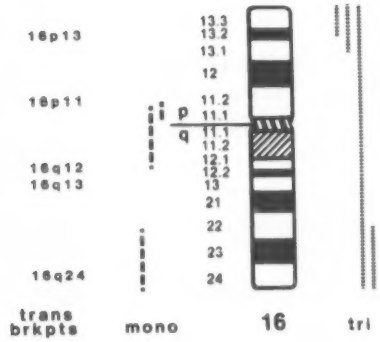
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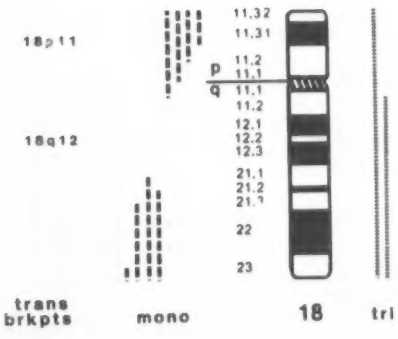
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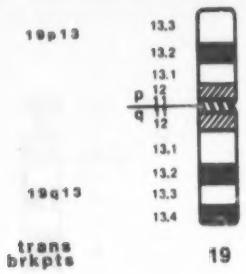
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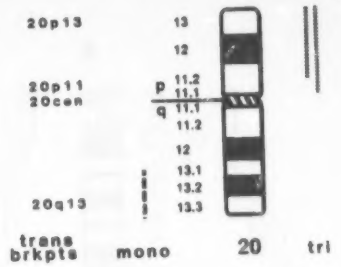
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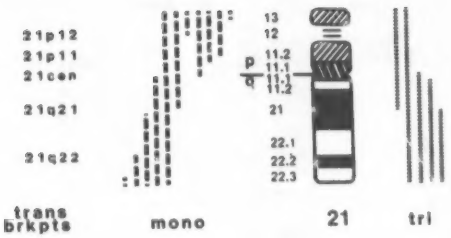
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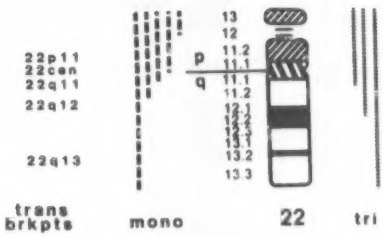
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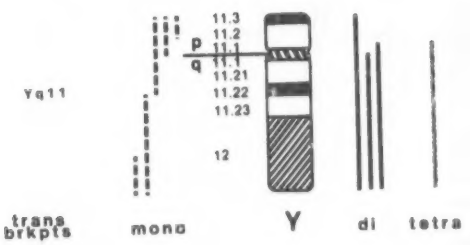
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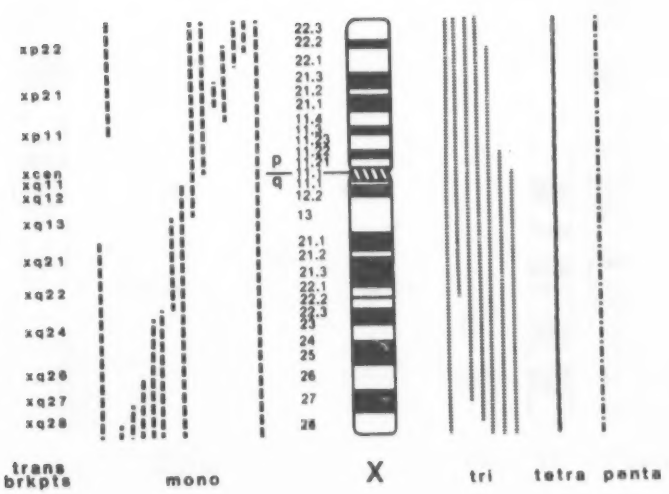
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penta

CHROMOSOME 1

Translocation Breakpoints

1p36.1	GM0126, t(1;15)
1p36	GM1230, t(1;2) GM3137, t(1;12) GM3423, t(1;6) GM4500, t(1;3)
1p34	GM1356, t(1;7) GM3501, t(1;8)
1p32	GM0201, t(1;17) GM4300, t(1;2)
1p31	GM1881, t(1;21)
1p13	GM3876, t(1;20)
1q12	GM0097, t(X;1) GM1813, t(1;21) GM7457, t(1;21)
1q23	GM4618, t(X;1)
1q32	GM0257, t(1;2) GM1550, t(1;3;4) GM3647, t(1;6) GM3648, t(1;6) GM4088, t(1;4) GM5348, t(1;9)
1q32.1	GM6924, t(1;3)
1q42	GM1421, t(1;6)
1q43	GM7212, t(1;6)
1q44	GM1564, t(1;21) GM3370, t(1;10) GM6227, t(1;16)

Monosomic Regions

1pter>1p36	GM1229
1p32>1p22	GM0605
1q21>1q25	GM2025
1q21>1q23	GM0803
1q23>1q32	GM2906
1q23.3>1q25.3	GM6038
1q25>1q32	GM0214 GM4940
1q41>1q43	GM5302
1q42>1qter	GM3126 GM6473
1q44>1qter	GM1564 GM3369 GM6226

Trisomic Regions

1q25>1qter	GM2836
1q25>1q32	GM0860
1q31>1q32	GM2946
1q32>1qter	GM3647 GM3648 GM5347
1q32.1>1qter	GM7158

CHROMOSOME 2

Translocation Breakpoints

2p25	GM1064, t(2;4) GM4012, t(2;5)
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2p13	GM1579, t(2;13)
2p11	GM4613, t(2;11)
2q11	GM2658, t(2;6) GM4300, t(1;2)
2q13	GM0327, t(2;8)
2q14	GM4045, t(2;4)
2q14.2	GM5010, t(X;2;8) GM5011, t(X;2;8)
2q31	GM1230, t(1;2) GM4139, t(2;13)
2q37	GM0693, t(2;21) GM0845, t(2;8) GM0846, t(2;8) GM3559, t(2;15)

Monosomic Regions

2pter>2p25	GM0501 GM2848 GM4371
2pter>2p24	GM0945
2pter>2p23	GM1138
2q23>2q32.2	GM3125
2q31>2q33	GM3918
2q37>2qter	GM0692 GM7158

Trisomic Regions

2pter>2qter	GM3576 GM4513
2pter>2p23	GM1848 GM1849 GM4409
2p23>2qter	GM4513
2q31>2qter	GM1229
2q33>2qter	GM2848

CHROMOSOME 3

Translocation Breakpoints

3p25.1	GM6924, t(1;3)
3p21	GM1550, t(1;3;4) GM2808, t(3;17) GM2963, t(3;17)
3p12	GM0439, t(X;3) GM1533, t(X;3)
3q13.2	GM2860, t(3;16)
3q13.3	GM4500, t(1;3)
3q21	GM0194, t(X;3) GM2899, t(X;3)
3q25	GM3882, t(3;10)
3q29	GM5564, t(3;9) GM3577, t(3;22)

CHROMOSOME 3 (Cont'd.)

Monosomic Regions

3pter>3p25	GM0366
	GM1253
	GM3039
	GM3085
	GM4627
	GM7873
3q29>3qter	GM3577

Trisomic Regions

3pter>3p21	GM4127
3pter>3p21.3	GM5336
	GM7216
3q12>3qter	GM3563
3q21>3q27	GM2486
3q21>3qter	GM0366
	GM3039
	GM3085
	GM4627
3q25>3qter	GM1253

CHROMOSOME 4

Translocation Breakpoints

4p16	GM0773,	t(4;7)
	GM1101,	t(4;12)
	GM4088,	t(1;4)
4p15	GM3811,	t(4;14)
	GM4921,	t(4;8)
4p14	GM1550,	t(1;3;4)
4q11	GM0624,	t(4;15)
4q21	GM1064,	t(2;4)
4q23	GM0380,	t(4;11)
	GM1561,	t(4;11)
4q25	GM1203,	t(4;12)
	GM4240,	t(4;6)
	GM5519,	t(4;11)
	GM5654,	t(4;11)
	GM7908,	t(4;14)
4q31	GM1063,	t(4;13)
	GM0972,	t(4;13)
4q31.1	GM4045,	t(2;4)
	GM4111,	t(4;5)
4q33	GM3886,	t(4;9)
4q34	GM3799,	t(4;11)
4q35	GM1091,	t(4;10)
	GM0157,	t(4;11)

Monosomic Regions

4pter>4p16	GM1220
4pter>4p16.1	GM1183
4pter>4p15	GM5336
4pter>4p15.1	GM4126
4pter>4p14	GM3435
	GM0072
	GM0343
4q31.1>4q31.3	GM4045
4q31.3>4qter	GM3013
	GM7267
4q33>4qter	GM2003
4q35>4qter	GM5401

Trisomic Regions

4pter>4p15	GM3810
4pter>4p11	GM0098
4q21>4qter	GM0501
4q31>4qter	GM4993

CHROMOSOME 5

Translocation Breakpoints

5p15	GM1678,	t(5;10)
	GM2426,	t(5;6)
	GM4111,	t(4;5)
5p14	GM0589,	t(5;14)
5p13	GM2487,	t(5;15)
5q22	GM3314,	t(5;15)
	GM6214,	t(5;11)
5q33	GM1536,	t(5;12)
5q33	GM3305,	t(5;9)
	GM4012,	t(2;5)
	GM5409,	t(5;17)
5q35	GM3494,	t(5;6)
	GM7173,	t(5;7)
5q35.3	GM5835,	t(X;5)

Monosomic Regions

5pter>5p14.1	GM2337
5pter>5p14	GM0344
	GM2800
	GM3912
5q35>5qter	GM3493

Trisomic Regions

5pter>p13	GM2488
5q33>5qter	GM1535
	GM4371

CHROMOSOME 6

Translocation Breakpoints

6p25	GM1137,	t(6;10)
	GM4240,	t(4;6)
6p23	GM1605,	t(6;11)
	GM3423,	t(1;6)
6p22	GM3494,	t(5;6)
6p21	GM1139,	t(6;13)
	GM2426,	t(5;6)
6q13	GM4188,	t(6;19)
	GM2594,	t(6;9)
6q15	GM2658,	t(2;6)
	GM7212,	t(1;6)
6q21	GM0610,	t(6;18)
	GM1421,	t(1;6)
	GM3404,	t(6;12)
	GM5183,	t(6;7)
	GM5184,	t(6;7)
6q22.2	GM5656,	t(6;21)

CHROMOSOME 6 (Cont'd.)

Translocation Breakpoints

6q25.3	GM7069,	t(6;10)
6q27	GM2068,	t(6;7)
	GM2669,	t(6;8)

Monosomic Regions

6pter>6p25	GM0144
	GM3647
	GM3648
6pter>6p24	GM6222
6q15>6q16.2	GM7212
6q25>6qter	GM6802

Trisomic Regions

6pter>6p22	GM3493
6q15>6q27	GM1221
	GM1222
	GM1524
6q22.2>6qter	GM5555

CHROMOSOME 7

Translocation Breakpoints

7p22	GM1696,	t(X;7)
7p15	GM0044,	t(7;10)
7p13	GM1356,	t(1;7)
7q21.2	GM5183,	t(6;7)
	GM5184,	t(6;7)
7q22	GM2068,	t(6;7)
	GM7173,	t(5;7)
7q32	GM0324,	t(X;7)
7q34	GM0657,	t(7;18)
	GM0773,	t(X;7)
7q36	GM4862,	t(7;21)
	GM5382,	t(7;18)

Monosomic Regions

7pter>7p22	GM1840
7p15>7p21.3	GM0044
7q22>7q32	GM1059
7q32>7qter	GM7412
7q34>7q36	GM0657
7q34>7qter	GM3240
	GM3733
7q36>7qter	GM7216

Trisomic Regions

7q34>7qter	GM1220
7pter>7p22.3	GM6097

CHROMOSOME 8

Translocation Breakpoints

8p23	GM6832,	t(8;15)
	GM0213,	t(8;12)
8p22	GM4921,	t(4;8)
8p21	GM1512,	t(8;10)
	GM3256,	t(8;15)
8p11.1	GM5010,	t(X;2;8)
	GM5011,	t(X;2;8)
8q11	GM1920,	t(8;14)
	GM1921,	t(8;14)
8q13	GM0845,	t(2;8)
	GM0846,	t(2;8)
8q21	GM2669,	t(6;8)
8q22	GM3501,	t(1;8)
8q24	GM0327,	t(2;8)

Monosomic Regions

8pter>8p21	GM1549
8q13>8q22	GM3134
8q24.1>8qter	GM3999

Trisomic Regions

8pter>8p21	GM3255
8pter>8p21.1	GM3689
8pter>8qter	GM0425
	GM0496
	GM2030
	GM2596
	GM4610

CHROMOSOME 9

Translocation Breakpoints

9p24	GM5348,	t(1;9)
	GM0344,	t(9;11)
	GM0705,	t(X;9)
	GM1751,	t(9;14)
	GM1892,	t(9;18)
	GM3305,	t(5;9)
9p22	GM6007,	t(X;9)
	GM5564,	t(3;9)
	GM2492,	t(9;13)
	GM2594,	t(6;9)
	GM3119,	t(9;17)
	GM3100,	t(9;17)
	GM3463,	t(9;14)
9p13	GM0988,	t(9;17)
	GM2821,	t(9;12)
	GM2822,	t(9;12)
	GM7114,	t(9;12)
	GM7548,	t(9;12)
9cen	GM3268,	t(9;21)
9q12	GM2836,	t(9;17)
	GM3886,	t(4;9)
9q13	GM1664,	t(9;13)
	GM6321,	t(9;22)
9q31.2	GM7456,	t(9;13)
9q34	GM1429,	t(X;9)

Monosomic Regions

9pter>9p24	GM3563
	GM5347

CHROMOSOME 9 (Cont'd.)

Monosomic Regions

9pter>9p22	GM1667
	GM2356
	GM5518
9pter>9p13	GM0870
9q11>9q21.3	GM1893

Trisomic Regions

9pter>9p24	GM1750
9pter>9p22	GM3462
9pter>9p13	GM2819
	GM2820
	GM6033
9pter>9cen	GM3268
9pter>9q11	GM3226
	GM5066
	GM5067
9pter>9q12	GM1734
9pter>9q22	GM1387
	GM1388
9pter>9q31.2	GM5319
9pter>9q34	GM1414
9pter>9qter	GM2329

CHROMOSOME 10

Translocation Breakpoints

10p15	GM1512,	t(8;10)
	GM3882,	t(3;10)
10p13	GM1678,	t(5;10)
10p11.2	GM6135,	t(10;21)
10p11	GM1137,	t(6;10)
	GM3370,	t(1;10)
10q11.2	GM7146,	t(X;10)
10q11.21	GM0044,	t(7;10)
10q22.3	GM5001,	t(10;17)
10q24	GM0216,	t(10;17)
	GM1091,	t(4;10)
10q26	GM1580,	t(10;21)
	GM1683,	t(2;10)
10q26.3	GM5878,	t(10;22)
10q26.1	GM7069,	t(6;10)

Monosomic Regions

10pter>10p15	GM3689
10pter>10p13	GM6936
	GM6937
10pter>10p11	GM3047
10p13>10p12	GM3470
10q26>10qter	GM1396
	GM1413
	GM1848
	GM1849
	GM3241
	GM4993
	GM5299

Trisomic Regions

10pter>10qter	GM1429
10pter>10p11	GM3369
10pter>10p11.2	GM6136
10q11.2>10q23.2	GM5047
10q21>10q24	GM5296
10q24>10qter	GM0217
	GM0959
	GM0983
10q26>10qter	GM1399
	GM1396
	GM1413

CHROMOSOME 11

Translocation Breakpoints

11p13	GM5519,	t(4;11)
	GM5654,	t(4;11)
	GM4613,	t(2;11)
	GM2859,	t(X;11)
11p11	GM3799,	t(4;11)
	GM6316,	t(11;16)
11p11.2	GM2859,	t(X;11)
	GM3316,	t(X;11)
11q11	GM0380,	t(4;11)
11q13	GM1695,	t(X;11)
	GM3005,	t(11;16)
	GM3322,	t(X;11)
11q23	GM0157,	t(4;11)
	GM0344,	t(9;11)
	GM1605,	t(6;11)
	GM3372,	t(11;22)
	GM3552,	t(X;11)
	GM3847,	t(11;22)
	GM4403,	t(11;22)
	GM7332,	t(11;22)
11q23.1	GM6214,	t(5;11)
11q23.3	GM6229,	t(11;22)

Monosomic Regions

11p14>11p13	GM6938
11p14.1>11p13	GM7736
11p13>11p11.1	GM5518
11p13>11p11	GM3118
	GM3808
11p13>11p11.2	GM2859
	GM3316
	GM6803
11q24>11qter	GM2008
11q25>11qter	GM0980

Trisomic Regions

11pter>p15	GM3435
11q23>11qter	GM0084
	GM3371
	GM4370
11q23.3>11qter	GM6228

CHROMOSOME 12

Translocation Breakpoints

12p13	GM1203,	t(4;12)
	GM3404,	t(6;12)
12p11	GM0213,	t(8;12)
	GM1101,	t(4;12)
	GM1665,	t(12;21)

CHROMOSOME 12 (Cont'd.)

Translocation Breakpoints

12q15	GM3137,	t(1;12)
12q24	GM1536,	t(5;12)
	GM2621,	t(X;12)
	GM2821,	t(9;12)
	GM2822,	t(9;12)
12q24.31	GM7114	t(9;12)
	GM7548	t(9;12)

Monosomic Regions

12pter>p13	GM4127
12pter>p11	GM0892
12q24>12qter	GM1535
	GM2819
	GM2820
12q24.31>12qter	GM6033

Trisomic Regions

12pter>12p11	GM1183
	GM0892
12pter>12qter	GM0892
	GM1990
12q24.31>12qter	GM7891

CHROMOSOME 13

Translocation Breakpoints

13p13	GM2927,	t(13;18)
13cen	GM0085,	t(13;13)
	GM0392,	t(13;22)
	GM1296,	t(13;15)
	GM2813,	t(13;15)
	GM3044,	t(13;14)
	GM4510,	t(13;14)
13q12	GM1664,	t(9;13)
	GM2971,	t(X;13)
13q12.1	GM7147,	t(13;16)
13q14	GM0972,	t(4;13)
	GM1063,	t(4;13)
13q14	GM2492,	t(9;13)
13q22	GM0627,	t(13;22)
13q32	GM1579,	t(2;13)
	GM4139,	t(2;13)
13q32.1	GM7456,	t(9;13)
13q34	GM1139,	t(6;13)

Monosomic Regions

13pter>13cen	GM1261
	GM2421
	GM6873
13pter>13q12	GM1387
	GM1388
	GM1840
	GM2018
	GM3241
13q12.3.13q21.2	GM2718
	GM7890
13q14.1>13q21.2	GM5877
	GM7312
13q14.1>13q22.1	GM1142
	GM1484
13q14>13qter	GM0509
13q22>13q32	GM2719
13q22>13qter	GM1570
	GM3518
	GM3887
13q34>13qter	GM1224
	GM6526
	GM2018

Trisomic Regions

13pter>13p11	GM1570
13pter>13q11	GM3518
13pter>13q14	GM1555
13pter>13q22	GM7150
13cen>13qter	GM0085
	GM2813
	GM6873
13q12.3>13q22	GM5750
13q12>13qter	GM0477
13q14.1>13qter	GM5832
13q14>13qter	GM1663
13q31>13qter	GM3089
13q32.1>13qter	GM5319
13pter>13qter	GM0503
	GM0526
13pter>13qter	GM2948
	GM3084
	GM3330

CHROMOSOME 14

Translocation Breakpoints

14p12	GM2044,	t(14;16)
	GM3811,	t(4;14)
14p11.2	GM0982,	t(14;20)
14cen	GM0005,	t(14;22)
	GM0479,	t(14;15)
	GM3044,	t(13;14)
	GM3417,	t(14;21)
	GM4510,	t(13;14)
14q11	GM1920,	t(8;14)
	GM1921,	t(8;14)
14q13	GM3463,	t(9;14)
14q21	GM0589,	t(5;14)
	GM1751,	t(9;14)
14q24.3	GM7912,	t(14;17)
14q32.1	GM0073,	t(X;14)
14q32	GM4410,	t(2;14)
	GM7908,	t(4;14)

Monosomic Regions

14pter>14p12	GM2044
	GM3810
	GM4610
14pter>14p11.2	GM0981
	GM5133
14pter>cen	GM3417
14q32>14qter	GM4409

Trisomic Regions

14pter>14q13	GM3462
14pter>14q21	GM1750
14pter>14q32	GM0074

CHROMOSOME 15

Translocation Breakpoints

15p11	GM0624,	t(4;15)
	GM2487,	t(5;15)
15cen	GM0479,	t(14;15)
	GM1296,	t(13;15)
	GM2813,	t(13;15)
15q11.2	GM0126,	t(1;15)
15q13	GM1813,	t(X;15)
15q15	GM1139,	t(15;17)
	GM4125,	t(15;18)
15q22	GM3559,	t(2;15)
15q25	GM6832,	t(8;15)
15q26	GM3256,	t(8;15)
	GM3314,	t(5;15)

Monosomic Regions

15pter>15p1	GM0118
15pter>15q11	GM0344
	GM1734
15pter>15q13	GM0017
15q26>15qter	GM3255

Trisomic Regions

15p11.2>15q13	GM2729
15pter>15qter	GM0682
	GM3133
	GM3184
	GM7189

Tetrasomic Regions

15pter>15p11.2	GM2729
15pter>15q11	GM3666
15pter>15q13	GM6246

CHROMOSOME 16

Translocation Breakpoints

16p13	GM2324,	t(16;22)
	GM6227,	t(1;16)
16p13.11	GM7147,	t(13;16)
16p11.1	GM6316,	t(11;16)
16p11	GM2044,	t(14;16)
	GM3005,	t(11;16)
16q12	GM2044,	t(16;18)
16q13	GM2860,	t(3;16)
16q24	GM3884,	t(X;16)

Monosomic Regions

16p11.2>16p11.1	GM5875
16p11>16q12	GM2044
16q22>16qter	GM2346

Trisomic Regions

16pter>16p13	GM2325
16pter>16p13.11	GM6226
16pter>16qter	GM4435
16q22>16qter	GM1396

CHROMOSOME 17

Translocation Breakpoints

17p13	GM0119,	t(17;22)
	GM0201,	t(1;17)
	GM0216,	t(10;17)
	GM1139,	t(15;17)
	GM2598,	t(Y;17)
	GM2808,	t(3;17)
	GM3196,	t(17;22)
	GM3197,	t(17;22)
17p13.1	GM7912,	t(14;17)
17p12	GM5409,	t(5;17)
17p11	GM3119,	t(9;17)
	GM3100,	t(9;17)
	GM2836,	t(9;17)
17q11.2	GM7151,	t(X;17)
17q12	GM6326,	t(Y;17)
17q23	GM0271,	t(17;19)
17q25	GM0988,	t(9;17)
	GM2963,	t(3;17)
	GM5001,	t(10;17)

Monosomic Regions

17pter>17p13	GM0217
	GM0959
	GM1663
17pter>17p13.2	GM6097

Trisomic Regions

17pter>17p13	GM1555
17q25>17qter	GM7150

CHROMOSOME 18

Translocation Breakpoints

18p11.3	GM4125,	t(15;18)
18p11	GM0610,	t(6;18)
18p11	GM2044,	t(16;18)
18q12	GM1892,	t(9;18)
18q12.2	GM2927,	t(13;18)
	GM5382,	t(7;18)
	GM0657,	t(7;18)

Monosomic Regions

18pter>18p11	GM2044
	GM3767
18pter>18p11.2	GM1727
18pter>18cen	GM1261
18pter>18q11	GM0892
18q21>18qter	GM2054
18q21.1>18qter	GM2980
18q21.3>18qter	GM0477

CHROMOSOME 18 (Cont'd.)

Monosomic Regions

18q23>18qter	GM0017
18q23>18qter	GM3089
	GM5832

Trisomic Regions

18pter>18qter	GM0143
	GM0734
	GM1359
	GM2422
	GM2732
	GM3250
	GM3538
	GM3623
	GM3769
18q11>18qter	GM1224

CHROMOSOME 19

Translocation Breakpoints

19p13	GM0271,	t(17;19)
19p13.3	GM4188,	t(6;19)
19q13.3	GM0089,	t(X;19)

Monosomic Regions

none

Trisomic Regions

none

CHROMOSOME 20

Translocation Breakpoints

20p13	GM1225,	t(2;20)
20p11.1	GM0982,	t(14;20)
20cen	GM7792,	t(X;20)
20q13.3	GM3876,	t(1;20)

Monosomic Regions

20q13.1>20qter	GM7945

Trisomic Regions

20pter>20p11.1	GM0981
	GM5133
20pter>20q11.2	GM2944

CHROMOSOME 21

Translocation Breakpoints

21p12	GM1700,	t(21.22)
	GM6943,	t(21;22)
	GM6944,	t(21;22)
21p11.2	GM5656,	t(6;21)
21p11	GM1665,	t(12;21)
21cen	GM2058,	t(21;21)
	GM3268,	t(9;21)
	GM3417,	t(14;21)
21q21	GM0693,	t(2;21)
21q21.2	GM1580,	t(10;21)
	GM4862,	t(7;21)
21q22	GM1813,	t(1;21)
	GM1881,	t(1;21)
21q22.3	GM6135,	t(10;21)
	GM7457,	t(1;21)

Monosomic Regions

21pter>21p13	GM4610
21pter>21p11.2	GM5655
21pter>21p11	GM4111
21pter>21cen	GM3268
21pter>21p12	GM2524
21pter>21q21.2	GM0692
21pter>21qter	GM0137
	GM0230
	GM1201
21cen>21qter	GM3417
21q21>21qter	GM2524
21q22>21qter	GM0983
21q22.3>21qter	GM6136

Trisomic Regions

21pter>21q21.2	GM1399
21pter>21qter	GM0201
	GM0260
	GM1137
	GM1920
	GM1921
	GM2067
	GM2419
	GM2524
	GM2571
	GM2767
	GM3576
	GM3606
	GM3716
	GM4435
	GM4592
	GM4614
	GM4616
	GM4617
	GM4965
	GM4927
	GM4928
21cen>21qter	GM0260
21q11>21qter	GM0144
21q21.2>21qter	GM1413

CHROMOSOME 22

Translocation Breakpoints

22p11	GM4628,	t(X;22)
22cen	GM0005,	t(14;22)
	GM0392,	t(13;22)
22q11	GM0119,	t(17;22)
	GM3196,	t(17;22)
	GM3197,	t(17;22)
	GM3372,	t(11;22)
	GM3847,	t(11;22)
	GM6229,	t(11;22)
	GM4403,	t(11;22)
22q11.1	GM1700,	t(21;22)
	GM6943,	t(21;22)

CHROMOSOME 22 (Cont'd.)

Translocation Breakpoints

22q11.1	GM6944,	t(21;22)
22q11.2	GM5878,	t(10;22)
	GM7332,	t(11;22)
22q12	GM2324,	t(16;22)
22q13	GM0627,	t(13;22)
22q13.3	GM6321,	t(9;22)

Monosomic Regions

22pter>22p12	GM4514
	GM5396
22pter>22cen	GM0392
22pter>22q11	GM3577
	GM5401
	GM0980
22pter>22q11.2	GM2944
	GM6526
	GM7149
22pter>22qter	GM2184

Trisomic Regions

22pter>22q11	GM0084
	GM3371
	GM4370
	GM6228
22pter>22q12	GM2325
22pter>22qter	GM7106

X CHROMOSOME

Translocation Breakpoints

Xp22.33	GM7146,	t(X;10)
Xp22	GM2971,	t(X;13)
Xp22.1	GM4618,	t(X;1)
Xp21.2	GM1695,	t(X;11)
	GM5835,	t(X;15)
	GM6007,	t(X;9)
Xp11	GM1813,	t(X;15)
Xcen	GM7792,	t(X;20)
Xq11.1	GM2859,	t(X;11)
	GM3316,	t(X;11)
Xq11.2	GM7151,	t(X;17)
Xq12	GM0705,	t(X;9)
	GM4628,	t(X;22)
Xq13	GM0073,	t(X;14)
	GM1429,	t(X;9)
Xq21	GM1696,	t(X;7)
Xq21.2	GM5010,	t(X;2;8)
	GM5011,	t(X;2;8)
Xq22	GM0089,	t(X;19)
	GM2469,	t(X;Y)
	GM2621,	t(X;12)
	GM3322,	t(X;11)
Xq24	GM0324,	t(X;7)
Xq26	GM0439,	t(X;3)
	GM1533,	t(X;3)
	GM3552,	t(X;11)
	GM3884,	t(X;16)
Xq27	GM0097,	t(X;1)
Xq28	GM0194,	t(X;3)
	GM2899,	t(X;3)

Monosomic Regions

Xpter>Xqter	GM0314
	GM0339
	GM0562
	GM0563
	GM0775
	GM0857
	GM0993
	GM1176
	GM1441
	GM1723
	GM1941
	GM3543
	GM4430
Xpter>Xp22	GM0314
Xpter>Xp22	GM4430
Xpter>Xp22.1	GM7149
Xpter>Xp11	GM0339
	GM0735
	GM5396
Xp22.2>Xp11.4	GM2470
Xp21.3>Xp21.1	GM7773
Xpter>Xcen	GM0088
	GM3543
Xpter>Xq13	GM1414
Xq11>Xqter	GM1941
	GM2103
Xq13>Xq22	GM3923
Xq21.1>Xqter	GM3827
	GM3828
Xq22>Xqter	GM2469
	GM4514
	GM6960
Xq22.3>Xqter	GM7148
Xq26>Xqter	GM3624
Xq27>Xqter	GM3935
Xq28>Xqter	GM7503
	GM7213

Trisomic Regions

Xpter>Xqter	GM0254
	GM3416
	GM3623
	GM3699
	GM4626
Xpter>Xq22	GM6960
Xpter>Xq27	GM3935
Xpter>Xq28	GM7213
Xp22>Xqter	GM0314
	GM4430
Xp11>Xqter	GM0735
	GM0339
Xcen>Xqter	GM0088
	GM2595
	GM3543

Tetrasomic Regions

Xpter>Xqter	GM0157
	GM0326
	GM1202
	GM1415
	GM1416

Pentasomic Regions

Xpter>Xqter	GM5009

Y CHROMOSOME

Translocation Breakpoints

Yq11	GM2469,	t(X;Y)
	GM2598,	t(Y;17)
Yq11.21	GM6326,	t(Y;17)

Monosomic Regions

Ypter>Yp11	GM6967
Ypter>Ycen	GM1709
Ypter>Yq11	GM0118
	GM2103
	GM2469
Yq11>Yqter	GM2730
	GM3595
Yq12>Yqter	GM2668

Disomic Regions

Ypter>Yqter	GM1250
	GM1993
	GM2521
	GM2587
	GM4375
Ycen>Yqter	GM1709
Yp11>Yqter	GM6967

Quadrisomic Regions

Yp11>Yqter	GM6967

APPENDIX E

AMISH MAJOR AFFECTIVE DISORDERS PEDIGREES

The Amish Study of major affective disorders (bipolar and unipolar forms) began in 1976 with grant support from the National Institutes of Mental Health. It was facilitated by the principal investigator (JAE) having observed and studied the health of this population over 25 years. Since 1976, cases of mental illness have been ascertained and multigenerational pedigrees based on bipolar probands have been documented clinically. Blood samples were collected for biochemical and genetic marker purposes from those pedigrees most informative for bipolar affective disorders.

Unique Research Setting

Amish isolates are well known for their genetic and cultural homogeneity and their value in medical genetic and molecular biologic study. They are recognized for their large families, clear paternity, geographic containment and extensive genealogic records. The Old Order Amish of Lancaster County in Pennsylvania constitute the oldest, or original group in the United States. This settlement was founded in pre-Revolutionary days by a small number of Swiss immigrants and has remained essentially a closed, endogamous group numbering some 12,500. As an in-bred population, this represents a rich resource for genetic studies of rare recessive disorders, common dominant conditions, chromosomal aberrations, the role of assortative mating, consanguinity and gene-environment interactions. Recently, Amish pedigrees have been cited as excellent general reference cell collections for characterization of new genetic probes and the construction of the human genetic map.

Psychiatric Diagnosis

Reliable diagnosis is crucial to a psychiatric genetic study. Careful diagnosis assures accurate clinical descriptions. All patients and their relatives were diagnosed by a five member psychiatric board using the latest, strict diagnostic criteria and uniform procedures for clinical assessment. Multiple sources of psychiatric information were obtained: direct interviews of patients and close family members about the patients, all available medical records, and continuing course of illness observations made biannually. High reliability of diagnoses has been demonstrated. The psychiatric status of individuals represented in the NIGMS Human Genetic Mutant Cell Repository is up-dated periodically.

Pedigree Selection

Pedigree 110 (IMR Family 884) was first identified as a key psychiatric resource at a workshop of NIMH genetic consultants in January of 1978. It was cited because of clustered psychopathology in large kinship lines with living grandparents and sibships of eight and more. Later it became one of a panel of five bipolar pedigrees used for extensive linkage studies. These pedigrees were segregating for affective disorders in a pattern that was potentially informative for genetic linkage.

The particular value of Pedigree 110 as a resource for other investigators led to developing permanent cell lines 1983. Lymphoblastoid and fibroblast cell lines were established for 51 individuals, with an additional 30 more family lymphoblastoid cell lines in 1984. Highest priority was shown those with confirmed major affective disorders and their "at risk" children and siblings. This increased the affective disorder cell collection to a total of 81 individuals: 23 with diagnoses and 58 well relatives. A companion pedigree (IMR Family 1075) linked to Family 884 by marriage and consanguineous ties, was introduced to the collection by tissue samples from an initial 12 persons, including patients with bipolar I, bipolar II and major depressive disorder.

Amish Study Data Base

A rich data base has been assembled for Amish pedigrees included in this catalog. Dr. Egeland, who serves as curator of the cell collection, has computerized data on selected biochemical variables and a number of conventional genetic markers typed in these pedigrees. Enough typing of these individuals has been done to demonstrate that considerable heterozygosity exists for both the classical markers (red cell antigens and serum proteins) and the DNA restriction fragment length polymorphisms (RFLPs) already typed by a network of Amish Study collaborators. The conventional genetic marker data are available on request by investigators ordering these cultures from the NIGMS Human Genetic Mutant Cell Repository. Scientists are encouraged to share the results of their linkage research. Dr. Egeland also will facilitate inquiries into the status of new DNA probes by referral to appropriate colleagues typing RFLPs in these pedigrees.

Futher Investigation

The intermarriage between IMR families 884 and 1075 provides numerous additional patients reaching a third and fourth generation. Each informative person added to such a kinship increases substantially the probability of linkage detection. Extending this valuable

national resource is highly desirable for both psychiatric linkage purposes and as general reference material. Psychiatric diagnostic evaluation is being done for all members of this multigenerational pedigree and plans are being made for general medical assessment of morbidity and mortality.

Inquiries should be addressed to:

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Dept. of Psychiatry (North Office)
University of Miami School of Medicine
49 Sylvania Road
Hershey, PA 17033

Office telephone: (717) 534-2038

Family 884 AMISH PRIMARY AFFECTIVE DISORDER PEDIGREE

GEN/LINE	ID	FIBROBLAST	LYMPHOBLAST	GEN/LINE	ID	FIBROBLAST	LYMPHOBLAST
I,18	118		GM7845	III,20	2105	GM5926	GM5927
II,02	157	GM5968	GM5969	III,21	2115	GM5928	GM5929
II,13	1114	GM5962A	GM5963B	III,23	2135	GM5897	GM5896
II,17	1154	GM5960	GM5961	III,24	2145	GM5899	GM5898
II,24	761	GM6014	GM6013B	III,25	771		GM7790A
II,25	762	GM6016	GM6015A	III,26	772		GM7813A
II,27	195		GM7815A	III,27	773		GM7812A
II,28	196		GM7789A	III,30	776		GM7814
II,29	197		GM7788	III,34	780		GM7816A
II,31	199		GM7840	III,83	680		GM7794
II,33	137		GM7836	IV,04	346	GM5906	GM5905
II,34	138		GM7943A	IV,05	356	GM5984	GM5983
II,37	141		GM7949B	IV,06	366	GM5976	GM5977
II,42	570		GM7779C	IV,07	376	GM5986	GM5985
II,43	571		GM7777	IV,08	386	GM5978	GM5979
II,44	572		GM7778C	IV,09	396	GM5980	GM5981
II,45	573		GM7776C	IV,17	741	GM5988	GM5987A
II,47	574		GM7948A	IV,18	742	GM5902	GM5901
II,49	575		GM7837	IV,19	743	GM5919	GM5918
II,50	576		GM7811	IV,20	744	GM5990	GM5989
II,61	119		GM7848A	IV,21	745	GM5908	GM5907
II,62	120		GM7847	IV,22	746	GM5915	GM5914
II,63	121		GM7846A	IV,23	747	GM6000	GM5999
III,03	235	GM5904A	GM5903	IV,24	748	GM5992	GM5991
III,04	158	GM5970	GM5971	IV,25	749	GM6002	GM6001
III,05	245	GM5972	GM5973	IV,26	740		GM5997
III,08	255	GM5974	GM5975A	IV,27	751	GM6004	GM6003
III,11	285	GM5994	GM5993	IV,28	752	GM6006	GM6005
III,12	295	GM5996	GM5995A	IV,29	271	GM5930	GM5931A
III,13	764	GM6024	GM6023	IV,30	272	GM5932	GM5933
III,14	765	GM6026	GM6025A	IV,31	273	GM5934	GM5935
III,16	767	GM6028	GM6027	IV,33	257	GM5917	GM5916
III,17	768	GM6018	GM6017	IV,34	276	GM5936	GM5937A
III,18	769	GM6030	GM6029B	IV,35	277	GM5938	GM5939
III,19	770	GM6012	GM6011	IV,36	278	GM5940	GM5941

AMISH PRIMARY AFFECTIVE DISORDER PEDIGREE

Family 884

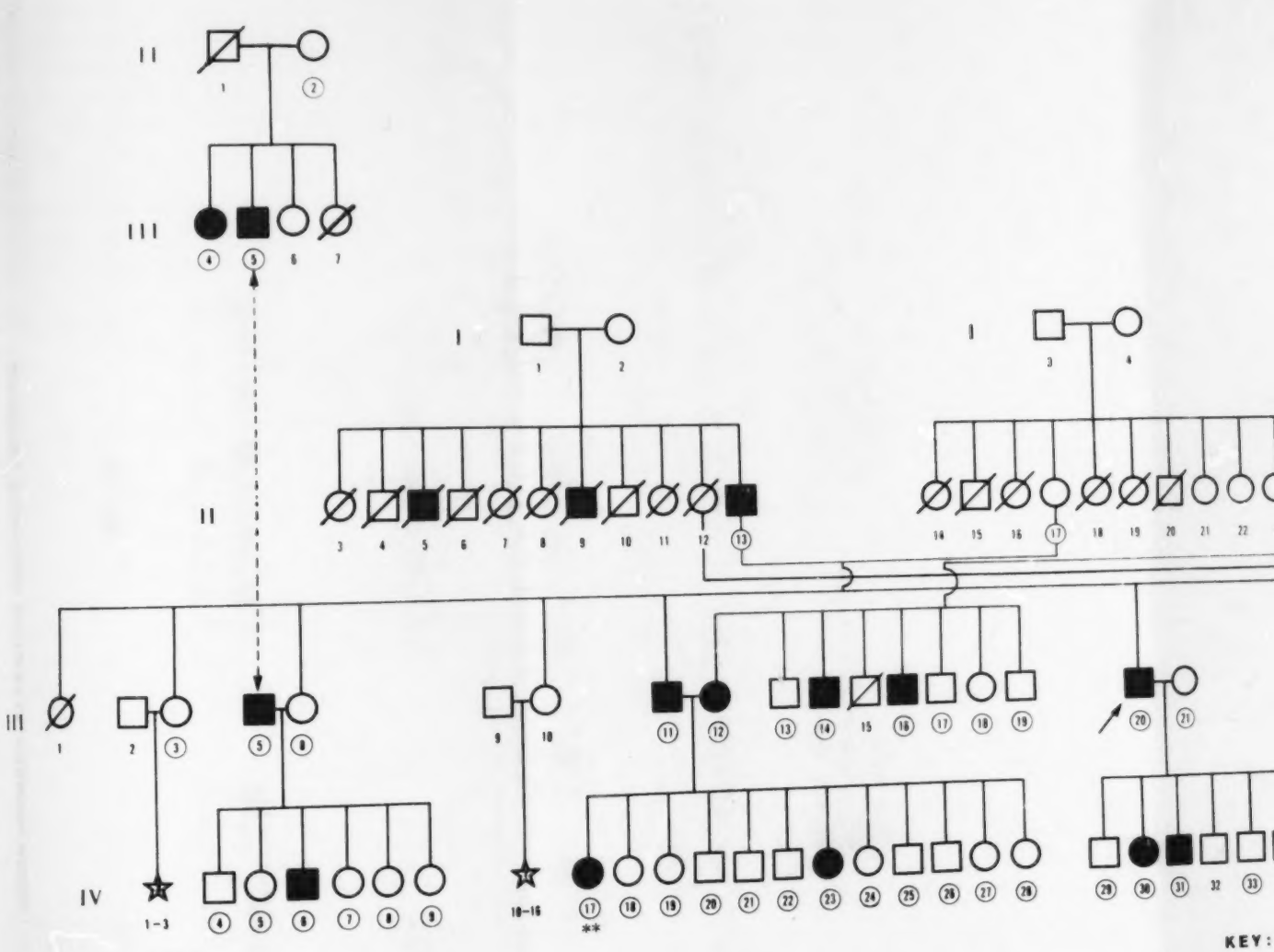
GEN/LINE	ID	FIBROBLAST	LYMPHOBLAST
IV,37	279		GM5943
IV,38	421	GM5895	GM5894
IV,39	422	GM5893	GM5892
IV,40	423	GM5889	GM5888
IV,41	424	GM5891	GM5890
IV,50	683		GM7798
IV,52	685		GM7944B
IV,53	686		GM7797
IV,54	687		GM7796
IV,56	689		GM7942
IV,57	690		GM7795

Family 1075

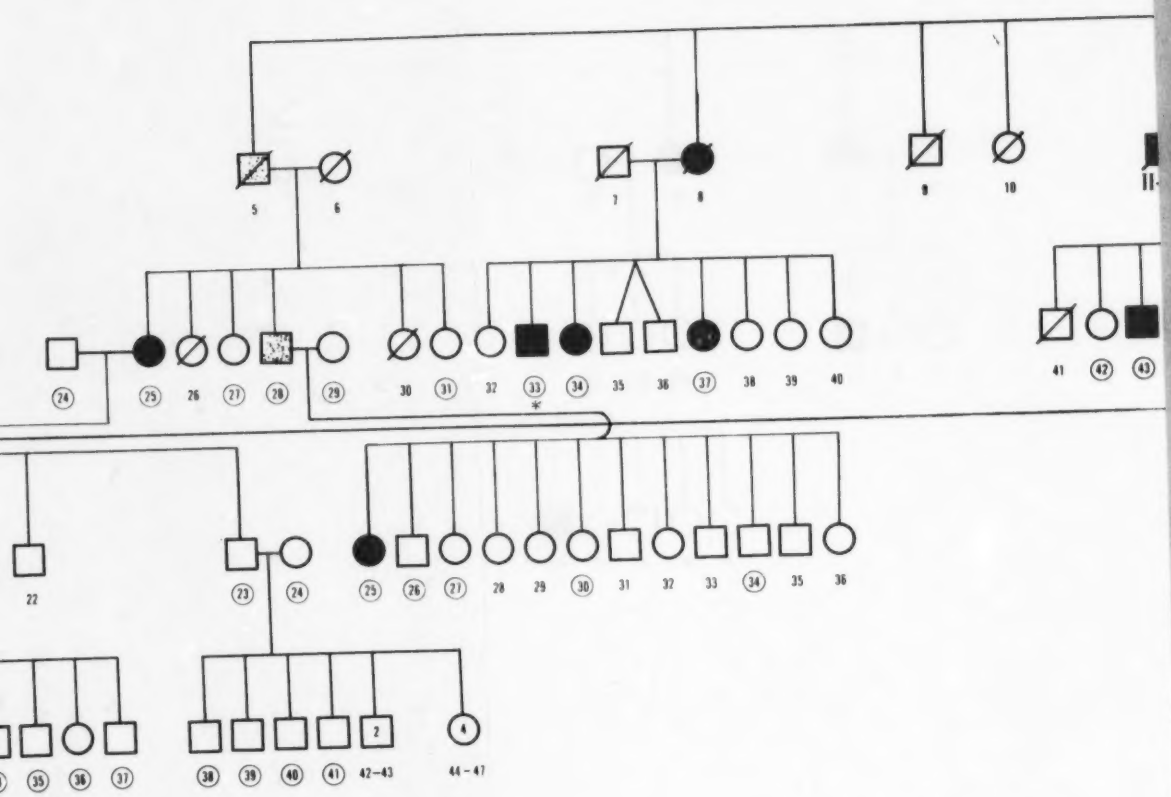
GEN/LINE	ID	FIBROBLAST	LYMPHOBLAST
III,04	910		GM7774
III,11	644		GM7775
III,15	645		GM7835
III,18	647		GM7849
IV,09	648		GM7784
IV,10	917		GM7785
IV,12	702		GM7874
IV,13	333		GM7875
V,23	651		GM7783
V,25	653		GM7787
V,26	654		GM7786
V,28	656		GM7876

FAMILY 884

AMISH PRIMARY AFFECTIVE DISO



DER PEDIGREE



AFFECTED

DECEASED

SEX UNSPECIFIED

SAME INDIVIDUAL

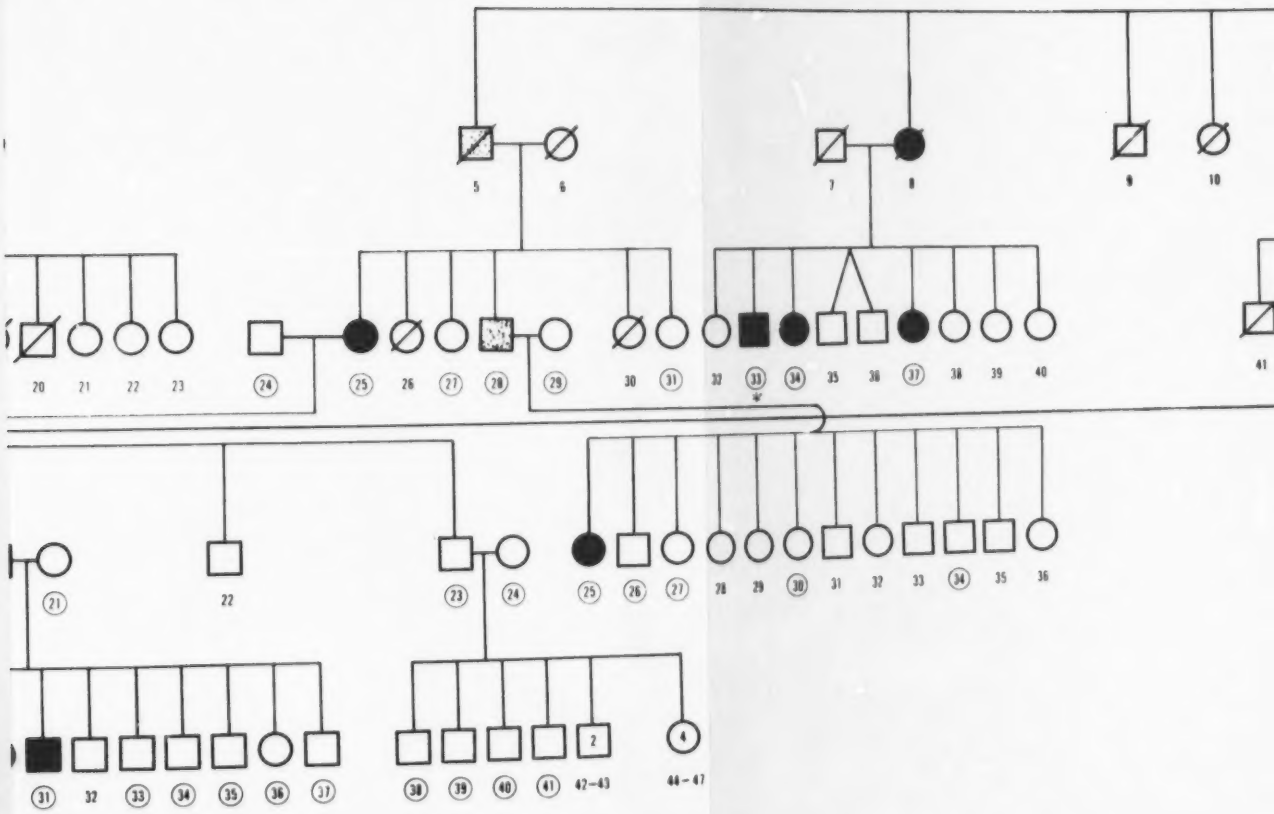
AVAILABLE FROM IMR - NIGMS

UNDIAGNOSED MENTAL ILLNESS

* FATHER OF IV-13 FAMILY 1075

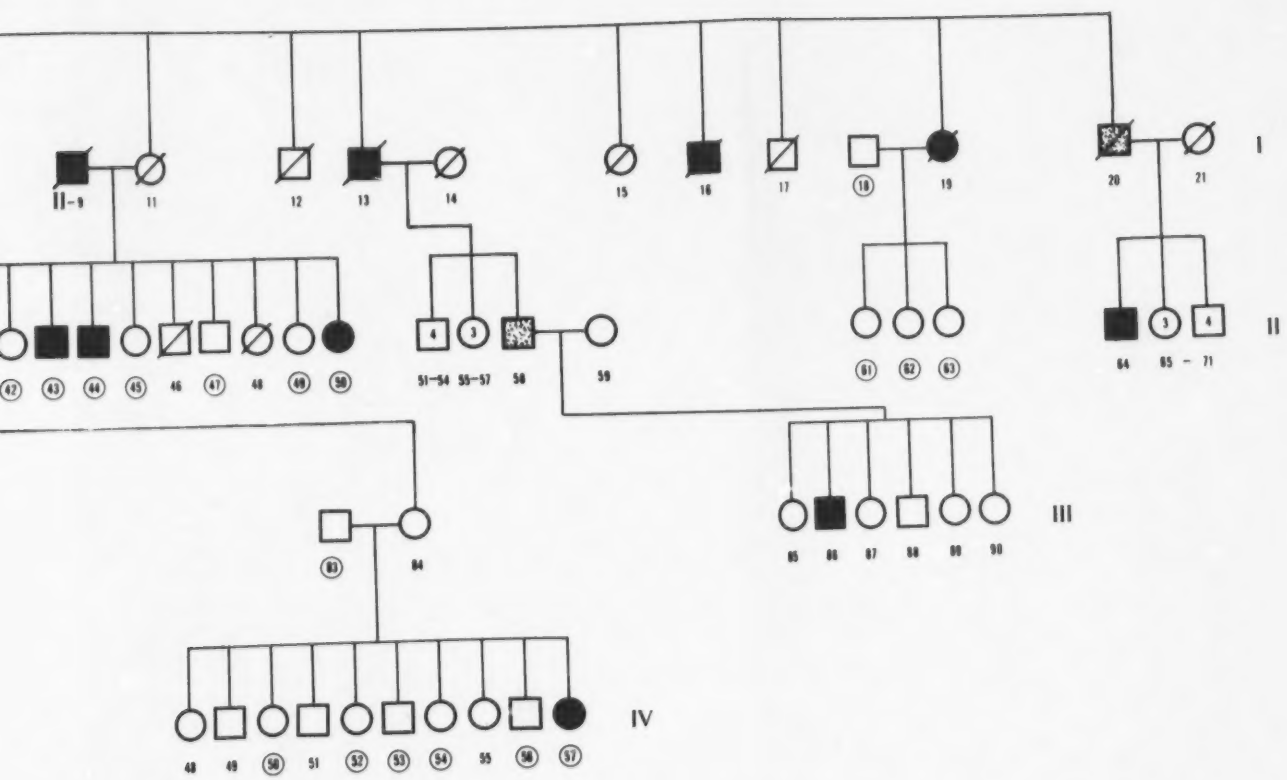
** WIFE OF V-28 FAMILY 1075

TIVE DISORDER PEDIGREE



KEY:

- AFFECTED
- DECEASED
- SEX UNSPECIFIED
- SAME INDIVIDUAL
- AVAILABLE FROM IMR - NIGMS
- UNDIAGNOSED MENTAL ILLNESS
- * FATHER OF IV-13 FAMILY 1075
- ** WIFE OF V-28 FAMILY 1075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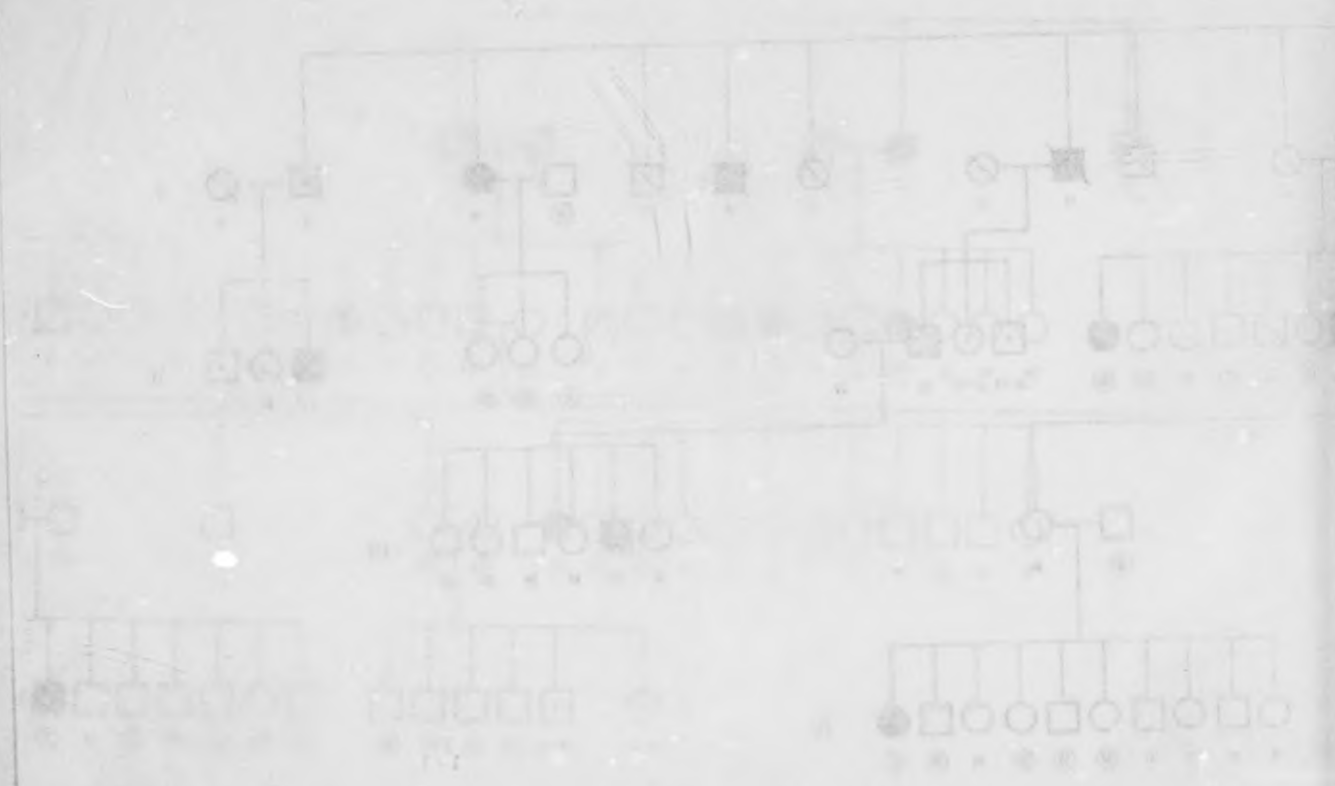
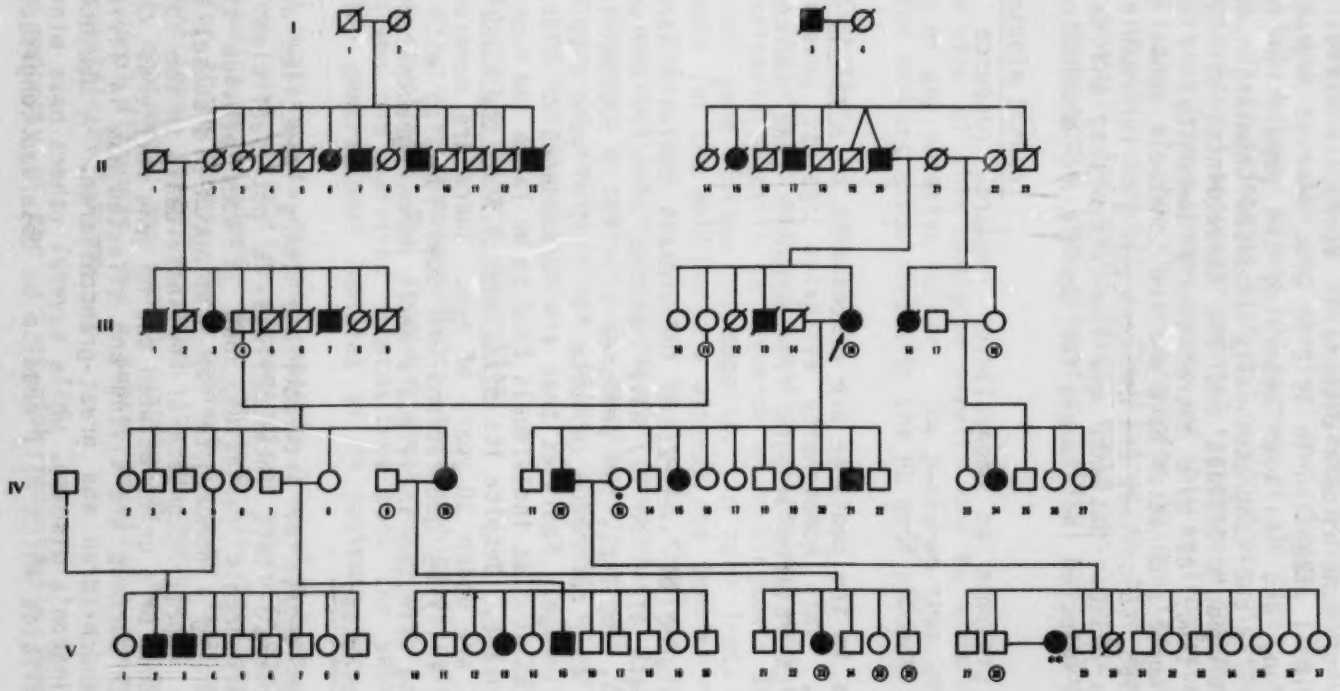


FAMILY 1075 AMISH PRIMARY AFFECTIVE DISORDER PEDIGREE



KEY:

- AFFECTED
- UNDIAGNOSED MENTAL ILLNESS
- DECEASED
- AVAILABLE FROM IMR-HIGMS
- * DAUGHTER OF II-23 FAMILY 884
- ** SAME INDIVIDUAL AS IV-17 FAMILY 884

APPENDIX F

VENEZUELAN HUNTINGTON'S DISEASE

AND REFERENCE COLLECTION

A unique focus of Huntington's disease in Venezuela was first brought to international attention in 1972 by Drs. Americo Negrette and Ramon Avila-Giron, who had been observing the population for twenty years. In 1977, the Congressionally mandated Commission for the Control of Huntington's Disease and Its Consequences strongly recommended further study of this extraordinary community. Field expeditions of one month's duration have visited Venezuela annually, from 1981 through 1983, to collect and document pedigree information, examine Huntington's disease patients and family members, and take tissue samples from selected individuals for genetic and biochemical research.

The community presents an unparalleled research resource in multiple ways:

Founder Effect:

All individuals in the pedigree are descendents of a single HD carrying progenitor. This homogeneity mitigates against possible confusion in research results arising from multiple gene sources.

Size:

The entire pedigree of over 2,600 individuals contains large nuclear families, each of which may have numerous affected and unaffected individuals. In fact, the pedigree size is so substantial that it is valuable as a reference resource for general gene mapping in addition to HD linkage. Families that are descendents of older, unaffected 50% and 25% at-risk individuals tend to be large and superb for general gene linkage. Despite its size, over 77% of the kindred are living and half are under 30 years of age. There are numerous instances of 3 and 4 living generations and some families with 2 generations of living affected individuals and a normal grandparent.

Possibility of Homozygotes:

There is a high degree of intermarriage among identified and potential gene carriers. This intermarriage is not sufficiently close to reduce polymorphic variation, but it does provide the possibility of producing a homozygote for Huntington's disease, if homozygosity is not lethal. Eighteen of the individuals in the collection are the product of a union between two HD gene carriers. One family in which both parents are living and affected has 14 living children and 56 grandchildren and great-grandchildren. A daughter definitely has Huntington's disease, while several others have minor neurological abnormalities which will need to be observed longitudi-

nally. The eldest is only 39 years old, so the majority are younger than the typical age of onset.

The Natural History of HD - no medications:

None of the HD patients sampled were taking any medications. All the families live in small villages along the shores of Lake Maracaibo and share the same physical environment. The climate is invariably hot, between 80 and 100 degrees at all times, with very high humidity. Their primarily fish and carbohydrate diet is standard and all other cultural and environmental factors are constant. Despite a shared HD gene from a common source and this environmental uniformity, the full range of phenotypic expression in Huntington's disease is observed in this community. The kindred is also large enough to provide juvenile and adult cases, rigid and choreic forms of the illness, monozygotic and dizygotic twins, and other subsamples.

The Venezuela Screen:

The size of this pedigree permits one to observe segregation patterns of any putative marker. The pedigree thus becomes a rapid screen for markers associated with the HD gene product.

Sample Selection:

The individuals selected for tissue sampling were chosen according to criteria specific to research on Huntington's disease linkage and general gene linkage and mapping. In many instances, families served both for the Huntington's disease and general reference collection. Collection rules were to sample complete sibships, both parents, and both maternal and paternal grandparents. If an individual was deceased or unavailable, samples were sought from that individual's parents and/or sibs. Often, a parent or grandparent had died of Huntington's disease.

The Huntington's Disease Collection:

Huntington's disease patients were the highest priority individuals for sampling. The second highest were their older at-risk siblings who had survived symptom-free past the years of highest risk. Two generational families of HD patients were also sought.

All Huntington's disease patients from whom tissues were taken, were given a quantitative, standardized neurological examination, many of which were filmed. Most patients have been examined sequentially for three years. Each patient is also assigned a Stage of Total Functional Capacity, which is the measure of their overall physical, emotional, and social functioning. They were assessed for other medical illnesses, and a subset received a neuropsychological and psychiatric evaluation.

The majority of the 50% at-risk individuals sampled also re-

ceived a quantitative neurological examination, particularly if on observation or from family report there appeared to be some physical or psychiatric problem. A portion of the at-risk individuals have been flagged as having minor neurological abnormalities. These individuals, in particular, will be assessed periodically to determine if these minimal abnormalities progress into Huntington's disease.

Reference Pedigree:

The first priority in collecting for the reference pedigree was to choose large sibships, with both parents and four grandparents living, if possible. In some instances, the reference families are identical to or built around those sampled for the HD collection. In other instances, these families are the offspring of at-risk individuals with a considerably reduced risk for Huntington's disease. If a marker is found to segregate with the HD gene, the reference families can be used as a genetically similar control group to check the specificity of the marker.

Paternity Confirmation:

All samples collected have been tested by a large panel of red cell and plasma markers to discover cases of non-paternity. No individual for which there is questionable paternity appears in this catalog pedigree. In some instances, HLA markers have also been determined.

Future Plans:

The pedigree published in these pages represents a fraction of a larger pedigree. The participants in the Huntington's disease project plan periodic visits to Venezuela to collect individuals whose addition could appreciably strengthen either the HD or reference material. Additionally, clinical follow-up is anticipated on all at-risk individuals sampled. The diagnosis of Huntington's disease is made conservatively, but if individuals can be definitively diagnosed and their genotype established, their tissue becomes substantially more informative. The Venezuelan Huntington's disease and reference collection is expected to increase in value with time and should serve as a major research resource.

Pooling Linkage Data:

The NIGMS Human Genetic Mutant Cell Repository is making these cultures available to investigators so that all can share a common source pedigree. The value of linkage research on this pedigree will be increased exponentially if scientists are willing to share the results of their linkage analyses. Pooling linkage data may reveal new linkages which otherwise would not be discovered by laboratories working independently. Dr. P. Michael Conneally, Director of the National Huntington' Disease Roster at the University of Indiana, has computerized the entire Venezuelan pedigree and has information regarding a number of conventional and DNA markers already available.

He has agreed to receive, coordinate and distribute linkage information in conjunction with his Roster functions to investigators willing to share data and enlarge the data base. Dr. Conneally can also be contacted regarding additional information about the family, including a more detailed pedigree with other family branches. He can be contacted at the following address:

Dr. P. Michael Conneally
Dept. of Medical Genetics
Indiana University Medical Center
1100 W. Michigan Ave.
Indianapolis, IN 46202
(317) 264-2241

List of Investigators:

The Venezuelan Huntington's Disease Gene Collaborative Project is an interdisciplinary effort by Venezuelan and US scientists to pursue the exceptional research potential of a Huntington's disease genetic focus in Venezuela. The following investigators have been associated with the Project:

Dr. Samuel Bailey
Dr. Ernesto Bonilla
Dr. F.J. Brinley, Jr.
Mr. Frederick A. Burnham
Dr. Michael Conneally
Dr. Richard Erbe
Dr. Jesus Esteves
Dr. Ramon Avilla Giron
Evelyn Goldstein, M.S.
Fidela Gomez, R.N.
Dr. Arthur E. Greene
Dr. James F. Gusella
Dr. Zulay Layrisse
Mr. Nelson Marsol
Dr. Ellen Messer
Dr. Adolfo Moreno
Dr. Humberto Moreno
Dr. Americo Negrette
Dr. John B. Penney, Jr.
Lennie Pireda, M.S.
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Dr. Marian Rivas
Ms. Greta C. Heider Rosenzweig
Dr. Ira Shoulson
Dr. Robert S. Snodgrass
Dr. Simon Starosta
Helen Travers, M.S.
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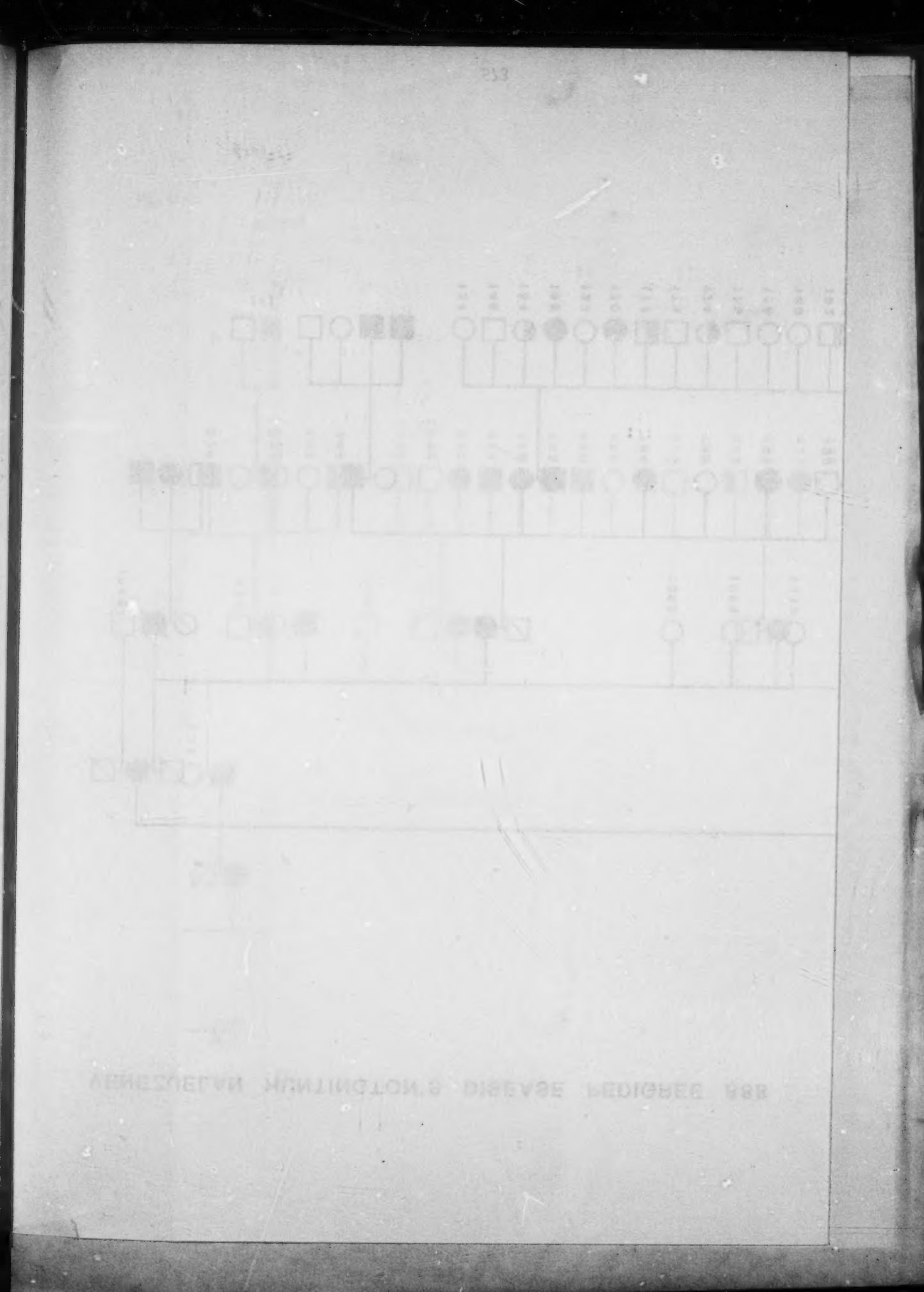
Dr. Alice R. Wexler
Dr. Nancy S. Wexler
Dr. Anne B. Young

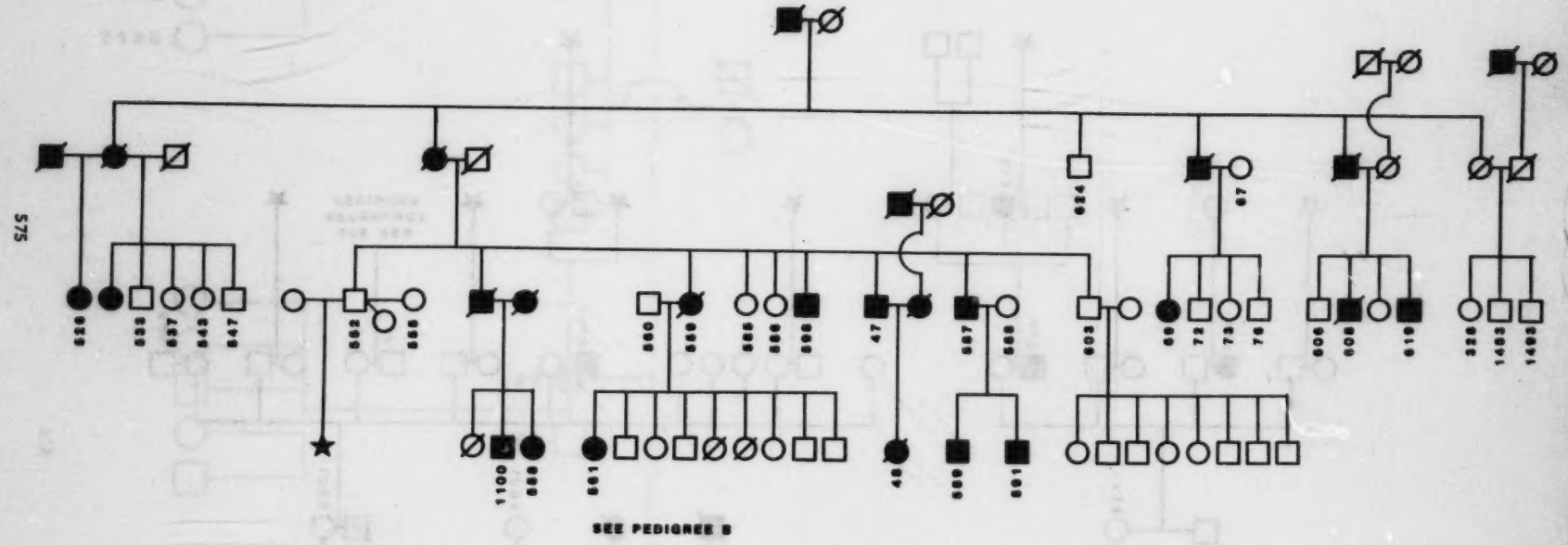
Charts Transposed to

End of Volume.

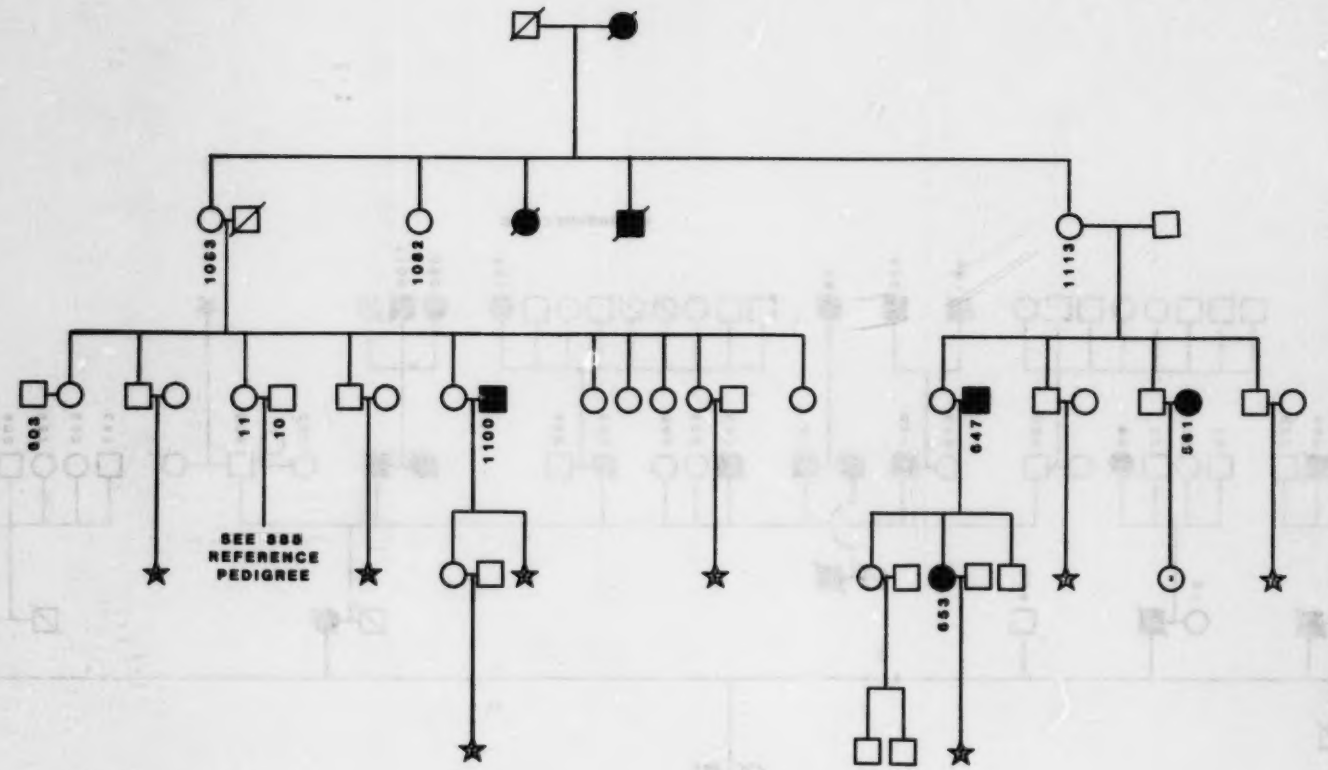


VENEZUELAN HUNTINGTON'S DISEASE PEDIGREE 934

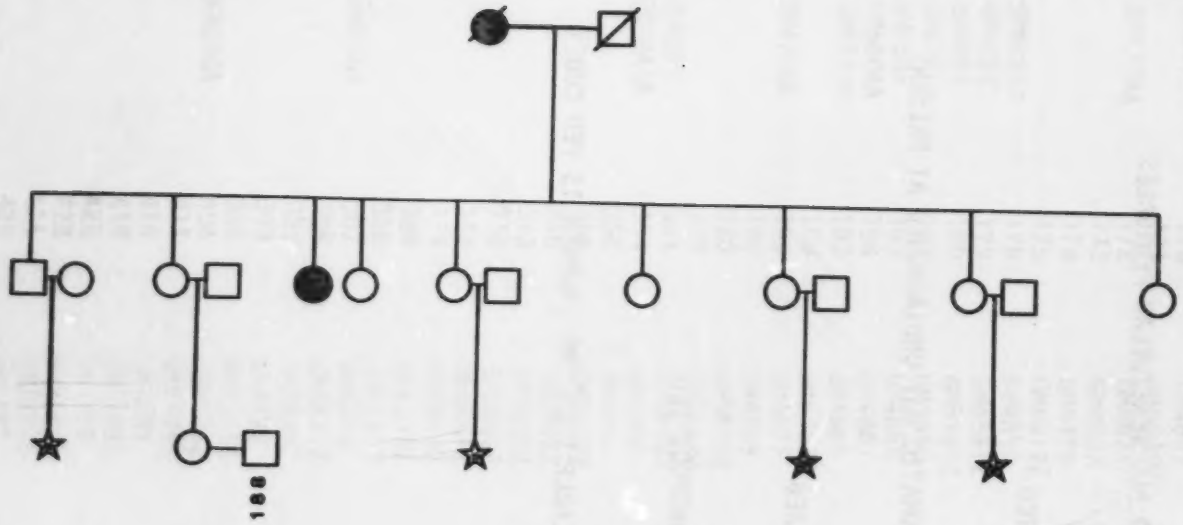
(SEE PEDIGREE B)



VENEZUELAN HUNTINGTON'S DISEASE PEDIGREE B



VENEZUELAN REFERENCE PEDIGREE A



KEY TO ALL VENEZUELAN PEDIGREES

AFFECTED

SYMPTOMATIC BUT UNDIAGNOSED AT PRESENT

DECEASED

SEX UNSPECIFIED

AVAILABLE FROM IMR (NUMBER IS PED CODE)

VENEZUELAN HUNTINGTON'S FAMILY ID LIST

Ped Code	Rep No Fibroblast	Rep No Lymphoblast	Ped Code	Rep No Fibroblast	Rep No Lymphoblast
7	GM5547Z	GM5546B	166	GM4849	GM5542B
8	GM4476	GM4477	167	GM6728Z	
9	GM4478	GM4479	170	GM4857	GM4856A
10	GM4711		171	GM6539Z	GM6538
11	GM4707A	GM4708A	172	GM4831	GM4830A
12	GM6397Z		173	GM4837	GM5376
13	GM6390Z		174	GM4829	GM4828A
15	GM5557Z		175	GM6512Z	
47	GM4867	GM4866A	178	GM4859	GM4858A
48	GM4281	GM4282	179	GM6541Z	GM5536B
67	GM6438Z	GM6437	180	GM5551Z	GM4290
69	GM4767	GM4766A	182	GM4853	GM4852A
70	GM6684Z	GM6685	183	GM4855	GM4854A
72	GM5603Z	GM5604A	184	GM4887	GM4886A
73	GM5571Z	GM5572A	187	GM4861	GM4860A
76	GM6780Z		188	GM6498Z	GM6499
119	GM4717	GM4718A	189	GM4877	GM4876A
120	GM6411Z		190	GM4865	GM4864A
121	GM6695Z		328	GM5699Z	GM5700A
122	GM6401Z		329	GM5649Z	
123	GM4482	GM4289	344	GM5609Z	GM5610A
134	GM4845	GM4844A	354	GM4809	GM4808A
135	GM6552Z		355	GM4801	GM4800A
136	GM6505Z		356	GM5679Z	GM5678A
137	GM6543Z		360	GM6456Z	
142	GM6510Z		373	GM5685Z	GM5684B
143	GM6550Z		374	GM5683Z	GM5682A
144	GM6549Z		378	GM5661Z	GM5660A
145	GM6548Z		379	GM5669Z	GM5668A
147	GM6542Z		385	GM6375Z	GM6376
148	GM6551Z		386	GM6369Z	GM6370
149	GM4847	GM4846A	387	GM4819	GM6348
150	GM6553Z		388	GM5641Z	GM5642A
151	GM6547Z		389	GM5625Z	GM5626A
152	GM6545Z		393	GM4727	
153	GM6546Z		396	GM5555Z	GM5554B
154	GM4689	GM4690A	404	GM5605Z	GM5606A
155	GM6467Z		411	GM6444Z	GM6443
156	GM6620Z		414	GM4693	GM4694
157	GM6461Z		419	GM5599Z	GM5600A
158	GM6468Z		420	GM4799	GM4798A
159	GM6404Z		423	GM5723Z	GM5724A
160	GM6410Z		424	GM5621Z	GM5622A
161	GM6472Z		425	GM4797	GM4796
162	GM6407Z		426	GM4795	GM4794A
163	GM6405Z		429	GM5744Z	
164	GM6399Z		441	GM4773	GM4772A
165	GM6409Z		442	GM4807	GM4284

VENEZUELAN HUNTINGTON'S FAMILY ID LIST

Ped Code	Rep No Fibroblast	Rep No Lymphoblast	Ped Code	Rep No Fibroblast	Rep No Lymphoblast
447	GM4703	GM4704A	659	GM4285	GM4286
450	GM5583Z	GM5584A	665	GM4729	GM4730A
453	GM4719	GM4720A	859	GM5627Z	GM5628A
456	GM4805	GM4804A	917	GM6776Z	GM6777
460	GM4787	GM4786A	920	GM6634Z	GM6633
463	GM4709	GM4710A	923	GM6656Z	GM6655A
469	GM4687	GM4688A	929	GM6766Z	GM6767
474	GM4715	GM4716A	930	GM6615Z	
480	GM6682Z		931	GM6652Z	GM6651A
482	GM5579Z	GM5580A	932	GM6448Z	GM6447
483	GM5589Z	GM5590A	933	GM6644Z	GM6643
526	GM5607Z	GM5608A	934	GM6632Z	GM6631
527	GM5593Z	GM5594A	940	GM6594Z	GM6593A
533	GM5447Z	GM5448	950	GM6582Z	GM6581
534	GM5613Z	GM5614A	963	GM5427Z	GM5428
537	GM5451Z	GM5452A	994	GM6586Z	GM6585A
543	GM5453Z	GM5454A	1002	GM6567Z	GM6566
547	GM5475Z	GM5476A	1010	GM5485Z	GM5486A
552	GM6480Z	GM6481	1040	GM6630Z	GM6629
555	GM5559	GM5558A	1068	GM6531Z	GM6530
558	GM4869	GM4868A	1069	GM6529Z	GM6528
559	GM5562Z	GM4818B	1070	GM6533Z	GM6532
560	GM4735	GM4736A	1080	GM6503Z	
561	GM4721	GM4722A	1081	GM6502Z	
585	GM6363Z	GM6364	1082	GM6628Z	
586	GM4745	GM4734A	1083	GM5691Z	GM5690A
587	GM4779		1086	GM6343Z	
588	GM4775		1087	GM6396Z	
589	GM4691	GM4692A	1088	GM6393Z	
591	GM6440Z	GM6439	1089	GM6392Z	
598	GM6772Z	GM6773	1090	GM6394Z	
603	GM6676Z	GM6677	1091	GM6386Z	
606	GM6365Z	GM6366	1092	GM6403Z	
608	GM4777	GM4776	1093	GM6470Z	
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612	GM4737	GM4738A	1095	GM6395Z	
613	GM6383Z	GM6384	1100	GM5689Z	GM5688A
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619		GM5560B	1192	GM6782Z	
624	GM4781	GM4780A	1195	GM5495Z	GM5496A
633	GM4791	GM4790A	1196	GM6768Z	GM6769
634	GM4815	GM4814A	1248	GM5739Z	GM5740A
640	GM4793	GM4792A	1249	GM5639Z	GM5640A
647	GM4287	GM4288	1483	GM6422Z	GM6421
653	GM4723	GM4724	1493	GM6735Z	

VENEZUELAN HUNTINGTON'S FAMILY ID LIST

Ped Code	Rep No Fibroblast	Rep No Lymphoblast	Ped Code	Rep No Fibroblast	Rep No Lymphoblast
1541	GM6523Z		2116	GM6406Z	
1542	GM6504Z		2117	GM6402Z	
1543		GM5530A	2120	GM6414Z	
1549	GM6521Z		2269	GM6749Z	
1554	GM6507Z		2271	GM6737Z	
1560	GM6500Z		2272	GM6757Z	
1561	GM5535Z	GM5534A	2274	GM6753Z	
1562		GM5544B	2275	GM6760Z	
1564	GM5527Z	GM5526A	2992	GM6424Z	GM6423
1565	GM6522Z		2993	GM6602Z	GM6601
1566		GM5524A	2996	GM6450Z	GM6449
1567		GM5528A	2999	GM6446Z	GM6445
1568		GM5552			

Although the Venezuelan family has been assigned three family numbers in the body of the catalog and two pedigree numbers in the appendix, it is one family. The multiple numbers simply satisfy computer needs.

APPENDIX G

UTAH PEDIGREES

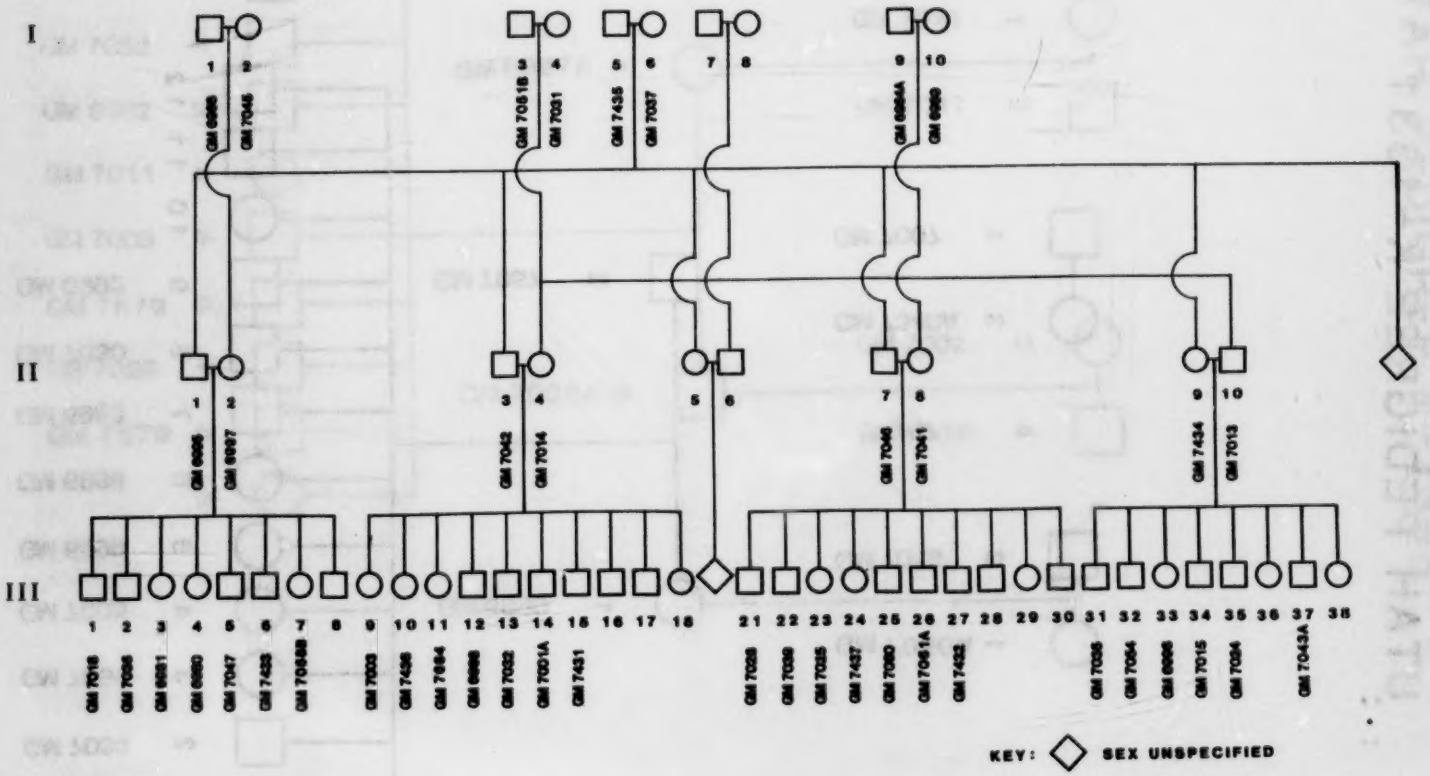
The following six extended-family pedigrees are intended to aid in the development of the human linkage map. Dr. Ray White is making these cultures available through the Human Genetic Mutant Cell Repository so that many investigators can share common source pedigrees.

The value of these pedigrees will be increased if scientists are willing to share the results of their linkage analyses. A data base of information pertaining to these pedigrees is maintained by Dr. White. He can be contracted at the following address:

Dr. Ray White
Howard Hughes Medical Institute
University of Utah Medical School
Salt Lake City, UT 84132

FAMILY 981

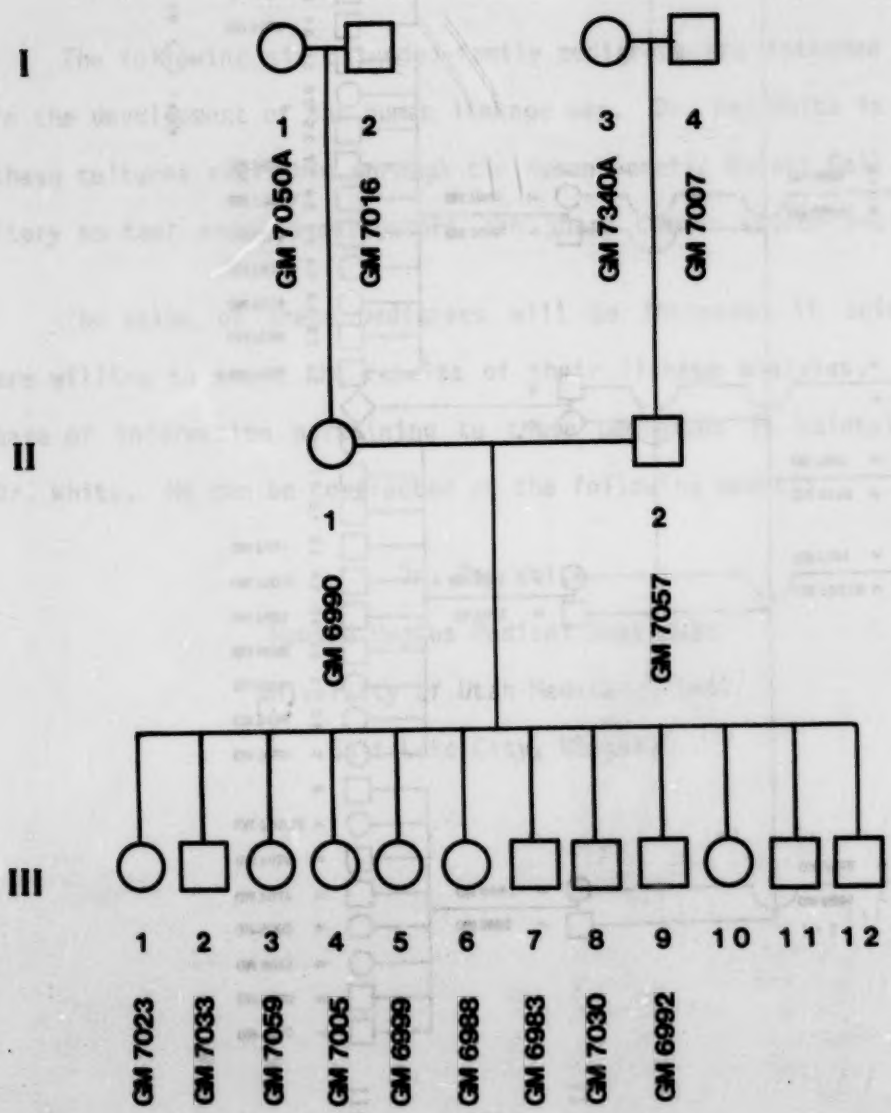
UTAH PEDIGREE K-1329



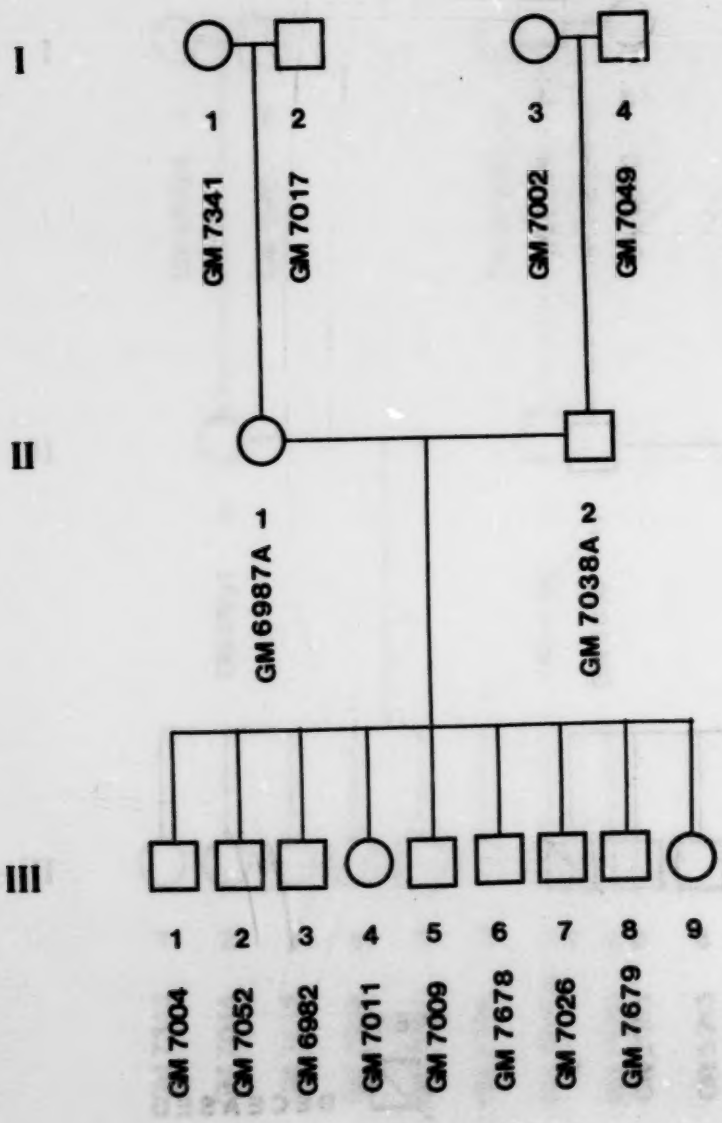
585

FAMILY 982

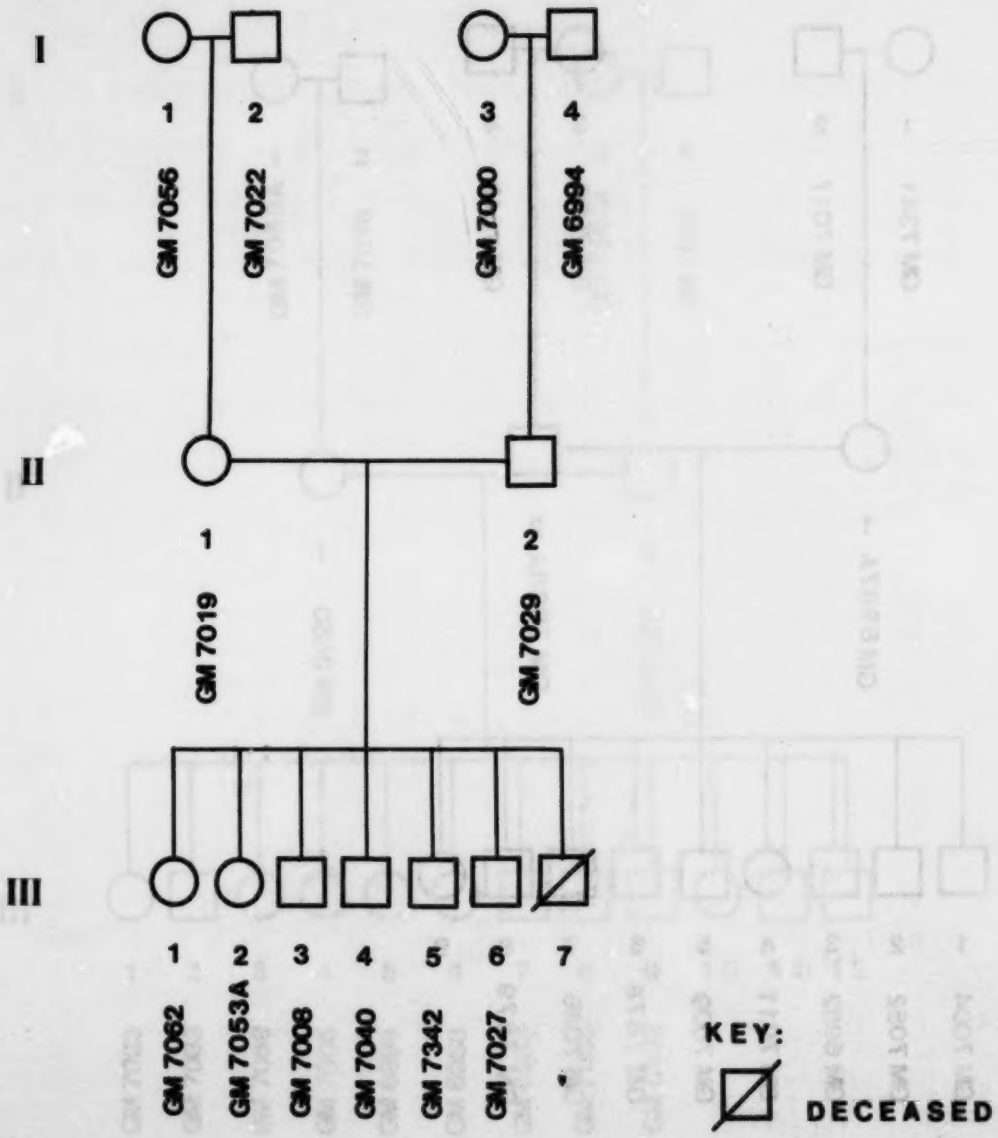
UTAH PEDIGREE K-1331



UTAH PEDIGREE K-1333

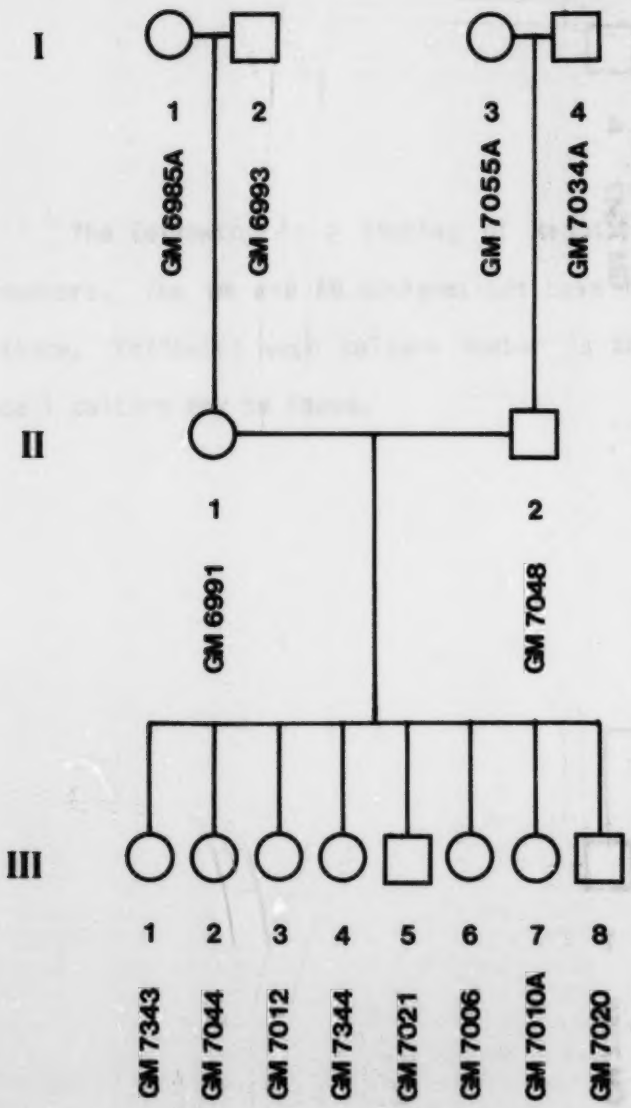


UTAH PEDIGREE K-1340



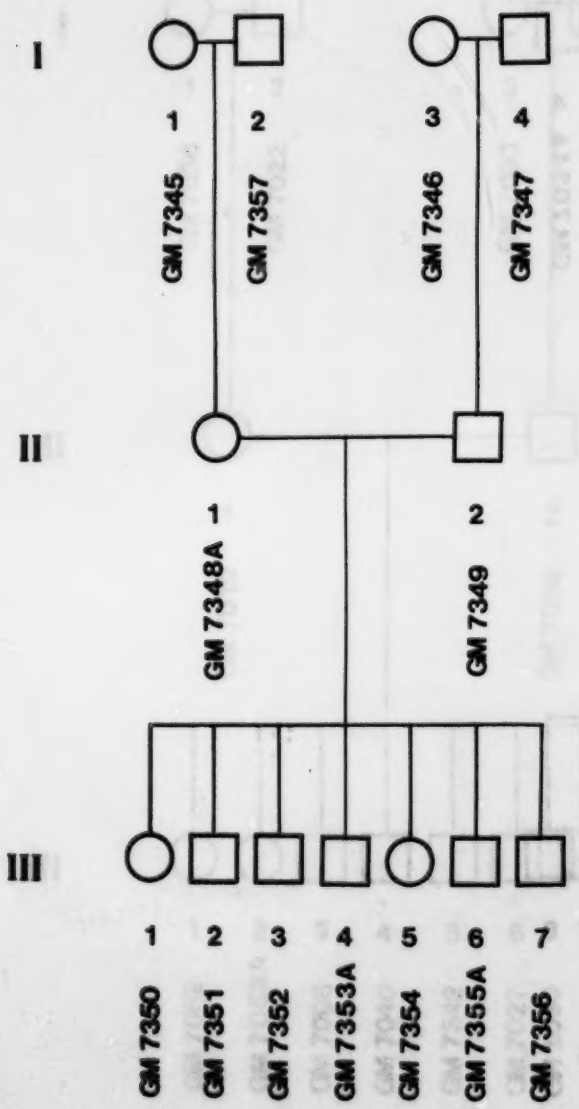
FAMILY 985

UTAH PEDIGREE K-1341



FAMILY 1029

UTAH PEDIGREE K-1345



APPENDIX H

Repository Number Versus Page Number Index

The following is a listing of Repository numbers versus page numbers. The GM and AG designations have been omitted for convenience. Following each culture number is the page(s) on which that cell culture may be found.

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APPENDIX I

ALPHABETICAL INDEX

The following is an alphabetical subject index. The numbers in parentheses following the titles refer to the McKusick catalog number. Diseases which have multiple names or the specific protein defect for a disease are cross-referenced utilizing the McKusick catalog number(s). Following each title and number are the page numbers in this catalog on which the title may be found. For example, Tay-Sachs disease (27277,27280), Beta-hexosaminidase A deficiency (27277,27280), and Gangliosidosis, GM2: Type 1 (27277,27280) are all referenced to the same Cell Repository catalog pages. Because space limitations do not permit all the various disease names or deficiencies for a given disorder to be listed in the text of the catalog, readers should look for the McKusick number in the disease heading on the appropriate catalog page.

In some cases, the number in parentheses following a title are preceded by the letters AG or GM. In these instances, these numbers represent Cell Repository identification numbers and not McKusick numbers. For example, the well known cell line DAUDI is followed by (GM3190) and the catalog page on which the culture GM3190 may be found.

*McKusick, Victor. Mendelian Inheritance in Man, 6th Edition. Johns Hopkins Press, Baltimore, 1983.

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Margin Index

To use, bend book in half and follow margin index to page with black edge marker.

NIGMS Human Genetic Mutant Cell Repository

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Biochemically Mutant Human Fibroblasts

Chromosomally Aberrant Human Fibroblasts

Virus-Transformed Fibroblast Cultures

Apparently Normal Human Lymphoblasts

Biochemically Mutant Human Lymphoblasts

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NIA Aging Cell Repository

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